

Consultation Response

Revision of the Directive 98/79/EC on *In Vitro* Diagnostic Medical Devices Response by European Genetic Alliances' Network

Introduction

1. The European Genetic Alliances' Network is an alliance of national genetic alliances and European disease specific patient groups with a special interest in genetics, genomics and biotechnology. We work for a voice in research and health policy and seek a world in which genetic diseases are understood, effectively treated, prevented and the affected people supported.
2. We welcome this review and the opportunity to respond.

Question 1:

- Would you consider the adoption of a risk-based classification for *in vitro* diagnostic medical devices as an improvement of the current European regulatory framework?
- Are you aware of any consequences for the protection of public health?
- Can you provide economic data linked to a change-over to this GHTF classification system?

3. European Genetic Alliances' Network 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.
4. 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.
5. A positive test for Huntington's disease for example, gives its recipient a certainty that he or she will suffer from this 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. (Though it should be recognised that the impact of an incorrect result for a genetic test for familial hypercholesterolaemia is severe, as the patient will not receive life saving medicine.)

Question 7:

Would it be necessary to maintain the exemption provided for by article 1(5) of Directive 98/79/EC and why?

6. 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.

7. In the UK there is a total of 498 genes for which tests have been authorised by the UK Genetic Testing Network (UKGTN www.ukgt.nhs.uk). There are a further 113 genes for which tests are undergoing assessment for authorisation, a number which reflects the likely growth of *in vitro* diagnostics in the genetic testing field.
8. 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.
9. It is vital that these tests continue to be available for use by health services. 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. The European Genetic Alliances’ Network believe that a continuation of the exemption in some form or another is therefore essential.
10. 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.

Question 8:

If the exemption provided for by article 1(5) of Directive 98/79/EC **should be clarified or limited**, which of the following items you would consider as appropriate in order to clarify the scope of this exemption and ensure a high level of safety:

Item 1:

Better define the concepts of “in-house test”, “health institution”, “premises of a manufacture or premises in the immediate vicinity”. Could you suggest an appropriate definition for these terms?

11. The European Genetic Alliances’ Network supports the proposal to better describe these concepts and supports a continuation of the exemption for those providing testing within the context of a health service. We do not believe ourselves to be best placed to advise on appropriate definitions, but expect any definition to include bodies providing tests as described in our answer to question 7.

Item 2:

Require that all “in-house tests” fulfil the **essential requirements** of the Directive 98/79/EC, **without being subject to a CE marking**?

12. No. The European Genetic Alliances’ Network rejects this proposal for the reasons stated in our answer to question 7.

Item 3:

Require that all **high risk** (*i.e.* class D according to GHTF classification) “in-house tests” are **excluded from the exemption** provided for by article 1(5) of Directive 98/79/EC and then have to fulfil the essential requirements of the Directive 98/79/EC including the involvement of a notified body?

13. No. As stated in our answer to question 1, many genetic tests should fall into a high risk category due to the potential impact that their results can have upon patients' and their families' lives.
14. As we have stated in our answer to question 7, any exclusion from the exemption will result in a lack of availability of valuable testing. This measure, to exclude high risk tests from the exemption, if applied to genetic tests, would severely restrict the availability of most of the most valuable genetic tests currently available.

Item 4:

Submit the health institutions and premises referred to in Article 1(5) of Directive 98/79/EC that manufacture "in house tests" to **accreditation**, based on ISO 15189, or **equivalent regulation** at national level?

15. 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.
16. The UKGTN provides regulation of genetic testing in the UK in a proportionate cost-effective manner. The European Genetic Alliances' Network believes that any measures to oversee quality, efficacy and safety of in vitro diagnostics exempt from this directive should follow a similar pattern to the UKGTN.

Question 9:

If the exemption provided for by article 1(5) of Directive 98/79/EC should not be maintained, would you consider it necessary to exempt in vitro diagnostic medical devices intended for diagnosis and monitoring of diseases or conditions affecting not more than 5 in 10,000 persons in the European Union from the scope of the IVD Directive and, if yes, why?

17. This proposal, to alter the current exemption from its basis on the provider of the test, to the subject of the test, is in the view of the European Genetic Alliances' Network, inappropriate. This would create a two-tier system *within* both "in-house" test providers and commercial test providers; and potentially lead to differences in quality of care for patients with rare conditions from those with more common conditions.
18. These measures could damage the flexibility for "in-house" providers to design tests for patients with more common conditions, thereby limiting the scope of healthcare available from certain healthcare providers. The European Genetic Alliances' Network believes that the exemption should continue to be based on the provider of the test rather than the subject of the test.

Conclusion

19. 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.



President
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