

**Advisory Committee on Genetic Testing
Pre-Natal Genetic Testing
Consultation Document: The Response of the Genetic Interest
Group (GIG)**

1. GIG is the UK Alliance of support groups for individuals and families affected by genetic disorders. It represents over 130 independent organisations, all of whom have been alerted to the existence of the consultation document and its recommendations. The report has also been considered by the Trustee Board of GIG. This response takes account of comments received by the Group and it has the endorsement of the Group's Trustees.
2. GIG welcomes the publication of the consultative document. We believe that the document itself and the recommendations and conclusions that it contains will form a sound basis for the development of Pre-natal genetic testing services in the UK. The framework for service delivery that would result should the document be implemented would provide a proper basis for ensuring the development of adequate, appropriate and accessible services throughout the UK for those who might wish to benefit from them and we would urge that the actions necessary to implement the report's recommendations should be put in place as soon as possible.
3. Having made the above point, there are a number of specific comments that we would make with regard to individual recommendations outlined within this report.
4. We are anxious that the issue of incapacity to consent should not be decided too precipitously. Work by groups such as "People First" or "Change" has shown that adults with learning disabilities are able to make sophisticated decisions for themselves if the information is presented in an accessible and appropriate manner. Many of GIG's own member groups have addressed the issue of discussing complex questions about genetic risk with children and in families and again, the overall view seems to be that children are capable of a greater degree of understanding than might be thought. Clearly a decision will have to be made on a case by case basis, but we would argue against a too easy assumption of responsibility by the doctor as the ultimate arbiter of what is, or is not in the patient's best interest. Wherever possible, good practice should include consultation with the family of those unable to make informed decisions themselves - recognising that there is potential for conflicting interests in the views obtained.
5. Although in many cases the information that may potentially be revealed about the future health of the child by a genetic test may be substantial the pattern of service delivery should reflect the likely impact of an adverse diagnosis. Not all mutations result in serious future health consequences. Whilst it is entirely appropriate and necessary that those who are identified as being at risk of having a baby affected by a major single gene disorder such as Tay Sachs disease or muscular dystrophy should have access to expert help pre and post test, the model of service delivery appropriate in these high impact situations is not necessarily appropriate under other circumstances. We would argue for the development of a range of possible responses by ante-natal testing services that reflect the

- possible severity of the condition and the different circumstances of women receiving the diagnosis.
6. GIG endorses absolutely the need for testing laboratories to provide high quality services and for these to be audited and accredited by appropriate external validation bodies. We are not convinced that either ISO9002 or CPA in themselves are capable of meeting adequately the needs for appropriate regulations in the light of current and possible future technologies and we would urge that this issue be kept under review. In particular, GIG would wish to see standards developed that ensure that test results are accurate, dependable or appropriate whether entire genes are being scanned for a large number of possible mutations, or large numbers of samples are being screened for a limited range of mutations known to be associated with particular conditions.
 7. GIG would wish to see any potential for future research into causes and cures for genetic disorders exploited to the full. At the same time care must be taken not to expose those coming forward for genetic testing to the possibility of unfair discrimination or disadvantage. A consistent approach to the issue of confidentiality and disclosure must be developed and we would strongly suggest that there should be close collaboration between the DoH, the MRC, research bodies such as the Wellcome Trust, the Pharmaceutical Industry and patient groups in developing appropriate protocols and procedures to ensure that adequate safeguards are put in place that will respect individual and family rights whilst at the same time encouraging research and maximising the opportunity for effective treatment to be developed.
 8. Finally and most importantly, GIG would wish to emphasis the need for services to be properly resourced if they are to deliver high quality, appropriate and accessible support for individuals and families at risk from genetic disorders. This is crucial, given the very short time available for ante-natal testing if women are not to be at risk of additional adverse consequences as a result of delays in either making the offer of testing or in supporting her in making decisions based on the results. At present there is considerable variation in access to services across the UK. This situation is unjustifiable and it is one which we believe should be addressed urgently by the Human Genetics Commission, the Department of Health and the NHS Executive if people at risk are not to suffer unfair discrimination and disadvantage and as a result have to live with the consequences of a failure to provide services and support which respond to the entirely reasonable and legitimate expectations of those affected by, or at risk from, genetic disorders.

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Director
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