The Norwegian Society of Medical Genetics (NFMG)
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June 16th 2007

Eurogentest

Cfr: Recommandations for genetic counselling related to genetic testing (Draft 2)

We acknowledge the important work and the high quality of the draft. The distinction between predictive testing for highly penetrant monogenic disorders and susceptibility testing for factors involved in multifactorial disorders is important and well described. It is an issue to become of major importance in the years ahead.

We support the draft, but have two comments that you may consider:

1) Cfr 6. General recommandations for genetic counselling, first section.

Genetic counselling should be mandatory when doing predictive testing for a highly penetrant and preventable inherited disorder, and the counseller should be obliged to inform about the preventive means and the right for the relatives to be informed.

> Arguments: It may be even more important with genetic counselling in preventable/curable inherited disorders, than in those which may not be prevented or treated. This is so because the preventable nature is an imperative to do something, and may induce reflections of guilt and responsibility if the consequences of a positive test is not properly explained and understood. This may be so both for the person tested, and for the relatives. In Norway, the legislation may be interpreted so that a consent to predictive testing includes the consent to receive the result, and it is codified that the result should only be given in the context of genetic counselling.
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> The post-test session should also include the offer of remittance to benefit from preventive means. Failure on delivering this information may induce ethical as well as legal questions on responsibility when a disease that could have been prevented, occurs. Both the ethical and legal arguments may also apply on the professional obligation to explain the need to inform the relatives of the availability of predictive tests leading to prevention for those at risk.

2) A practical description of what a high penetrant genetic defect is, and what a genetic susceptibility factor is, is difficult but may be necessary. We have suggested to our Government, that any single genetic factor having 50% or higher penetrance, should be considered a monogenic disorder, and genetic testing in healthy persons should be considered as predictive. Diseases with lower life-time penetrance than 50% may be specifically included due to the preventability or medical importance of the condition (e.g. long QT-syndrome).