Committee for Public Relations and Ethical Issues of the
German Society of Human Genetics

Statement on Population Screening for Heterozygotes

I. Advances in human molecular genetics have produced new possibilities to identify
heterozygotes (carriers) for recessive hereditary illnesses. The most important example
is with the demonstration of a mutation responsible for cystic fibrosis (mucoviscidosis).
About every 20th individual in our population is a carrier of this disease. This mutation
can be directly detected in about 75% of carriers in the Central European population.

Such tests will likely become available for other diseases in the future. With them, it will
be possible to determine the heterozygote status of healthy individuals in large
populations by population screening. For affected persons, the results of the
examinations may present an important new option for their life and family planning.
Therefore the development and utilization of corresponding tests is necessary for ethical
reasons.

However, the German Society of Human Genetics rejects population screening at
present, since the necessary framework is absent. This includes in particular an
adequately informed public, sufficient information about the acceptance or rejection of
such test procedures by the public, and the guarantee that the necessary genetic
counselling will be available.

II. Qualified information and individual genetic counselling in different phases of such a
test procedure are absolutely necessary for the following reasons:

1. Comprehensive information about the frequency, etiology, symptoms, course,
and therapy of the disease for which heterozygosity is to be determined is
indispensable. Only a fully informed individual is qualified to consent to or to
reject such an investigation. In addition, such information safeguards against
affected persons and their families facing discrimination.

2. When heterozygosity is demonstrated, comprehensive counselling about the
significance of the results is required, especially to prevent false judgements that
lead to stigmatization.
If two partners are heterozygotes, they should be informed of all their options,
which include accepting their existing genetic risk and, possibly, a child with the
disorder in question; abstaining from having their own children; adoption; and
prenatal diagnosis. Pressuring the couple, even indirectly, to utilize prenatal
diagnosis or to terminate a pregnancy must be avoided.

III. The following must be guaranteed in addition to individual genetic counselling before larger populations are systematically examined:

1. Comprehensive, qualified information of the population
   This must occur before a heterozygosity test is introduced on a wide scale. The danger of social pressure to utilize such test methods can be countered in this way.

2. Voluntary utilization
   Making the tests part of, e.g., routine obstetrical examinations or testing at the request of a third party (e.g., employer, insurance agency) must be precluded.

3. The ability of the proband to comprehend the importance of the decision
   The person to be tested as a rule should be of legal age.

4. The qualification of genetic counsellors and laboratory personnel
   Adherence to the "Guidelines for Carrying Out Molecular Genetic Diagnostic Procedures" and "Basic Principles of Genetic Counseling" of the Berufsverbandes Medizinische Genetik [Professional Association of Medical Geneticists] is obligatory.

IV. The German Society of Human Genetics supports the position of the Berufsverbandes Medizinische Genetik on heterozygote screening for cystic fibrosis, especially their requisition that accompanying scientific pilot projects be introduced prior to each screening program.