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

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Statement on Genetic Diagnosis in Children and Adolescents

1. Genetic diagnosis in children and adolescents is indicated if it is necessary for the differential diagnosis of manifest symptoms or for establishing the etiology of a disease.

2. A predictive genetic diagnosis is indicated during childhood if the onset of a disorder can be regularly expected at this age and if medical measures can be taken to prevent the disease or its complications or to treat the disease.

3. As a rule, predictive genetic diagnosis should not be carried out in a healthy child for a disorder with onset in adulthood. An exception to this rule can be seen only when there is an established, effective, and important medical treatment that can be offered in case the test result is positive. Otherwise, respect for the child`s autonomy in deciding whether to utilize a genetic test takes precedence over the wish of third parties, including the parents, to carry out predictive diagnosis. This should always be delayed until the child is able to understand not only the genetic facts of the situation but also the emotional and social consequences of the various possible test results. As a rule, an individual can be expected to have this ability by age 18 years.

4. However, deferring a predictive genetic diagnostic test should not prevent discussing the disease in question with the child in a manner appropriate to his/her age, including how it is inherited and the possibility of its being diagnosed. If, for personal reasons, the parents press for their child to be tested for a disorder that normally becomes manifest in adulthood, the family should be fully advised about all conceivable aspects of the test. If the test is not indicated from a medical point of view and there are no other pressing reasons for carrying it out, it may be refused. The more severe the diagnosis or disorder in question, the more compelling the reasons must be for the investigation to be carried out.

5. For some diseases the available data may be inadequate or uncertain so that it cannot be decided with certainty may not be certain that a predictive diagnostic test would be medically significant for the (not yet) affected child. Before tests are introduced for such diseases, the problem of their medical usefulness and psychosocial significance must be investigated. In addition to medical evaluations, corresponding research projects should include psychosocial evaluations since the latter become even more important clinically if the purely medical benefits are uncertain or insignificant. For diseases in which the possible medical benefits of predictive diagnosis justify a study, the psychosocial

evaluation may provide valuable knowledge about possible psychosocial factors in predictive diagnostic tests for other disorders.

6. An investigation for the sole purpose of determining the carrier status for a recessively inherited illness or a balanced familial chromosomal translocation should not be carried out since the results would only be significant for future reproductive decisions of the child him/herself. Therefore the examination should be deferred until the child can understand all the associated facts and psychosocial implications and asks for the test him/herself. However, by consulting with the parents it should be established that they understand the significance of utilizing or not utilizing the test so that they can be responsible for the child`s being informed at a later date.

7. On occasion, knowing the carrier status of a child can contribute to the genetic counseling of other family members. Although children should not be routinely included in such studies, it may be that knowing a child`s carrier status would allow a more exact statement about the genetic status of family members who would like to have the information. Here, each case should be individually evaluated to determine whether the tests are necessary and how helpful possible results would be. Furthermore, it should be established that the child will be offered genetic counseling when he/she is older. The significance of these tests for the child and the family should be evaluated prospectively and retrospectively.

8. Information about the heterozygote status of the child may unavoidably become available from a prenatal investigation or from tests needed to diagnose unexplained signs and symptoms. The parents should be advised beforehand that information about the possible heterozygote status of the child will definitely be available at a later date. However, as a rule such a finding should not be communicated for the time being if it is not absolutely necessary for dealing with the problem at hand. In such a situation, safeguarding the child`s right to determine independently whether he/she wants the information may conflict with the parent`s desire for information. Since the conflicts of all possible case constellations can not covered by a single regulation, the decision about how to deal with information about a child`s heterozygote status should be worked out in each case by the persons involved.

9. In connection with an adoption the results of a child`s genetic tests could influence decisions made for the child. However, a genetic test should not be a requirement for giving a child up for adoption or for finding it a suitable home. Recommendations 1 to 7 are also valid here. Each individual case should be discussed with a medical geneticist, and all relevant factors considered before decisions are made.

10. The above principles are valid for genetic diagnosis in adolescents under the age of 18, as for that in children. However, adolescents should be included in the consultations and decision-making process in a manner appropriate for their degree of maturity and state of development.

Reference: Kommission für Öffentlichkeitsarbeit der Deutschen Gesellschaft für Humangenetik e.V. (1995) Stellungnahme zur genetischen

Diagnostik bei Kindern und Jugendlichen. Med Genet 7:358-359