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Legal issues of genome analysis

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Abstract: A number of legal issues arise with regard to genome analyses. In the context of medical treatment, some of these issues are being dealt with by the German Gendiagnostikgesetz. In the context of science, the right to self-determination over personal data should be observed. A legislative act would be necessary to create a reliable basis for scientists, doctors, and patients.

Keywords: freedom of research; genetic data bank; German gene diagnostics act; incidental finding; informed consent; right to free development of personality and self-determination.

Legal issues of genome analysis

Genome analyses are now technically feasible and can provide a higher gain in knowledge for research and medicine. They also enable personalized medicine.

As there is no separate legal regime for genome analyses in Germany, it raises the question about the legal framework that does apply to such analyses. It is important to distinguish between the genome analysis for medical purposes – in this case, the Genetic Diagnostics Act applies (hereinafter, using the official German abbreviation: GenDG) – and the genome analysis for research purposes. In this case, the federal and state data protection laws, the right to freedom of research in accordance with Article 5(3) of the German Basic Law (hereinafter: GG) and the personal rights pursuant to Article 2(1) GG apply.

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1. The following is a brief overview of the essential requirements under the GenDG with respect to genome analyses. The GenDG contains both prerequisites for genetic testing and analysis and for the use of genetic samples as well as the handling of genetic data. However, the GenDG is limited to the application areas “medical purposes” and “the purpose of clarifying parentage”. In other words, genetic testing – and thus also genome analyses – for research purposes does not fall under the scope of the GenDG.

   However, once the GenDG applies, the essential requirements of the law, that is, testing limited to medical doctors (Section 7 GenDG), patient information (Section 9 GenDG), consent (Section 8 GenDG) and consultation (Section 10 GenDG) must be observed.

   The issues of patient information and consultation figure in genome analyses in a particular way; the contents of patient information under the GenDG include, among other things, also the nature, significance, scope, purpose, type, extent and meaning of the genetic test. In addition, the patient should also be informed about his or her options regarding treatment, prevention and avoidance. Moreover, the patient must be informed about his or her right not to know, as well as his/her right to revoke consent [1].

   It is obvious that patient information, as envisaged under the GenDG, is not practical or feasible in connection with a full genome analysis. After all, the patient would have to be fully informed about all genetic susceptibilities that may be discovered as part of a genome analysis. Keeping in mind that the human genome contains approximately 3.2 billion pieces of genetic information, comprehensive patient information, as provided for under the GenDG, is not practical or feasible.

   The right not to know is also difficult to enforce in this context: a genome analysis does not only reveal the information for which the genetic analysis was ordered, but also findings that should not be part of the actual test. These are so-called secondary findings. While the law itself does not include any

explicit provisions for this, the Genetic Diagnostics Committee's (hereinafter: GEKO) directive on the requirements of the contents of patient information in connection with genetic testing for medical purposes pursuant to Section 23(2)(3) GenDG has formulated the following specification: The patient must be informed about unexpected genetic characteristics and secondary findings (II.1 of the GEKO directive on the contents of patient information in connection with genetic testing for medical purposes pursuant to Section 23(2)(3) GenDG, as amended on 27.04.2012, amended on 16.11.2012, published and entered into force on 03.12.2012).

Under this GEKO directive, the affected person is to be informed that he/she can determine what is to be done with the secondary findings. Without any doubt, this probably refers also to knowledge about unexpected genetic characteristics. This GEKO specification for so-called secondary findings will likely apply especially when a test is done that is bound to reveal more findings than those actually pursued, as is the case with genome analyses.

2. Apart from this, there is the legal regime in place for testing for research purposes. There is no legal framework in this area that would be similar to the GenDG, although data protection provisions generally apply. However, this will involve a decision on the two basic rights concerned, that is, the right to freedom of research under Article 5(3) GG, according to which the arts, science, research and teaching are to be free, as well as the general personal right under Article 2(1) GG, which provides for the free development of one's personality, provided that he or she does not infringe the rights of others and does not contravene public policy and order or moral law.

   The right to informational self-determination and the right not to know are derived from the general personal right under Article 2(1) GG [2]. The required consent in connection with genetic testing and the option to revoke such consent are also products of the general personal right under Article 2 GG. Since neither of the two basic rights is expressly subject to a statutory reservation, one must employ practical concordance to determine how to apply both constitutionally guaranteed basic rights side by side. In the process, one must ensure that the core of the basic rights remains intact and is not unduly restricted.

   An essential element of such a joint application of both basic rights is the requirement of informed consent. According to this concept, the patient must give his/her consent freely after having been informed, which is to enable him/her to take an independent decision. As already explained above, this raises questions in connection with genome analyses and total sequencing. One question is: how can the patient be informed and advised, when the nature and scope of the expected findings cannot be fully estimated and mapped? Another question involves the further use of data for research purposes, as well as the one question that comes up in medical practice all the time regarding the subsequent sharing of information with test subjects or their family members. In answering these questions, one must make sure, above all, that the right to informational self-determination, including the right not to know, is satisfied.

   As for uses for research purposes, the legal assessment may well be relatively straightforward if all personal information is removed from the genetic data. However, this raises the question whether it is possible to anonymize genetic data completely. Under current German and European data protection laws, it is assumed that this is the case, because the issue has not yet been addressed more thoroughly and because the German Federal Data Protection Act defines that data are already considered anonymous if the person can be identified only with considerable effort, such as another genetic test [3].

   However, if the personal reference is to be maintained, it will be necessary to pseudonymize the data. It will also be necessary to put in place rules about patient information, consultation, the right not to know, and revocation of consent particularly in respect of new insights and new findings. Only if patients have made an informed decision, such consent may be permissible in connection with the sharing of personal data, thus providing a legal cover for researchers.

3. Another growing issue in the context of genome research concerns genetic databases. Genetic researchers have an interest in collecting findings from genome analyses in databases that are accessible internationally in order to expand the references for potential findings and insights. However, storing the data in such a database also requires the informed consent of the test subject or patient. Accordingly, the purpose of the database must be covered by the consent.

Finally, of course, attention must be paid to data protection laws, that is, the exchange of data must be transparent and, from the point of view of data protection

laws, secure. Generally, this can be achieved only through a complex system of pseudonymization – as is already being done in the case of biological databases in coordination with the respective Ethics boards and Committees.

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