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Direct-to-consumer Genome Tests on the Internet: Social, ethical, and legal dimensions, with special consideration of the Gene Technology Law (Gentechnikgesetz, GTG)

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Summary

Since autumn 2007, companies have been offering analyses of not only particular genetic variants but also much more comprehensive, so-called genome-wide analysis to consumers on the internet. For about 300-2,000 Euros (depending on the scope of services included) these companies analyse up to a million points (single nucleotide polymorphisms, SNP) across a person's genome and produce an individual genetic risk profile. This profile claims to convey indications as to what diseases and heritable conditions, as well as physical characteristics and character traits, customers are genetically predisposed to. The individual risk profile also entails information on drug metabolism; some companies also offer analyses of genetic ancestry (which were not the subject of this study).

The marketing and sale of these tests has been carried out mainly on the internet so far. Customers order a so-called spit kit which they fill with a saliva sample and return to the company by post. A few weeks later, they can access their test results online by means of a personal password.

Consultations with physicians, or genetic counselling by means of face-to-face encounters with genetic counsellors, are not typically part of this process.

This situation was the point of departure for this study. Since the beginning of the direct-to-consumer (DTC) marketing of genomewide tests on the internet – a phenomenon which has come to be known as personal genomics (PG) -, a dearth of literature discussing its likely societal, ethical, and legal implication has been published. This report provides a brief overview of the main issues raised in this literature. It then discusses existing legal provisions pertaining, and recent regulatory reactions, to the PG-market in three countries which of strategic importance for Austria: the US, the UK, and Germany.

At the core of the study then lies a detailed discussion of the practical questions raised by the test process and the test results for the tested individuals. This aspect has not been systematically examined in the past. For this reason, the author of this study underwent a PG test provided by the two commercial companies which, at the time of this study, offered the cheapest and yet most comprehensive tests. Both the testing process, as well as the test results, were described in detail; test results received from both companies were compared, and open questions were highlighted. Special consideration was given to the issue of clarity and accessibility of test results.

With regard to the two companies analysed in this study, the way that they convey test results to consumers are clear and accessible; the limited predictive value of the genetic markers tested are highlighted consistently, and the importance of non-genetic factors in the aetiology of many complex diseases (cardiovascular diseases, diabetes, arthritis, etc) are emphasised. Lifestyle and environmental aspects are portrayed as important factors with the ability to decrease or increase disease risk. In cases where customers have filled in questionnaires about lifestyle etc on the companies' websites, these factors are considered also in the interpretation of the DNA data. In sum, regarding the way in which test results are communicated to test-takers, it seems that companies pose considerable emphasis on the accessibility and educational value of the information. **However the author of this report deems problematic the great extent to which companies encourage consumers to share or upload personal information (medical histories, information pertaining to prescription as well as recreational drug use), without drawing attention to the risks inherent in sharing such information in the middle and in the long run**. In addition, the genetic counselling available through PG companies is limited to counselling via e-mail or over the phone and does typically not include possibilities for face-to-face encounters between counsellors and test-takers. Furthermore, language issues represent a further potential barrier to the

accessibility of genetic counselling offered by PG companies (it can be assumed that not everybody who is capable of reading and understanding their test results in English is also able to discuss these in English).

In the case of the author of this report, test results received from both companies (*23andMe*, and *Pathway Genomics*) were consistent. Both the list of diseases for which the author was diagnosed with an increased risk, as well as those for whom she was told to have a decreased risk, were largely identical between the two companies. Also with regard to drug metabolism and carrier status of heritable diseases, results from both companies were identical. This indicates that the clinical validity of the tested markers is high, and that both companies use similar algorithms for the calculation of individual risk. An aspect that remains problematic is the dubious clinical utility of test results: the levels of increased and decreased genetic risk communicated to individual consumers are so marginal that they lose their practical relevance. (In the case of the author, her 'increased risk' for heart disease consisted of 8.9% life time risk, compared to 7.4% life time risk prevalent in the general population, which amounts to a difference of 1.5%!). **Consequently, the risk attributed to PG-Tests in most of the existing literature, namely that they are likely to cause needless worries or groundless relief of health concerns, seems less realistic than the scenario that test-takers could turn to their family physicians for help with the interpretation of their test result. This would pose undue strains on the public health care system.**

Some –albeit not immediate – clinical utility could be seen in the effect of PG-test-results to instigate test-takers to adopt healthier lifestyles (more frequent exercise, healthier diets, etc). In the case of information on carrier status for heritable diseases, clinical utility can also manifest itself in test-takers being able to make more informed decisions and devise strategies for risk-reduction (eg having their partners tested etc). At the same time, however, PG tests also bear the risk of over-prevention, namely that test-takers seek for additional advice and/or to undergo preventive treatments for all conditions for which according to their PG-test they could be at higher genetic risk. Also this would mean a considerable additional burden on the health care system.

Finally, the report entails a detailed discussion of the question about the extent to which provisions of the Austrian Gene Technology Law (*Gentechnikgesetz*, GTG) are, or could be, applicable to the PG-market. The GTG was conceived and came into effect at a time when the current developments were not foreseeable. The author situates open questions with regard to two main aspects:

(1) The fact that the definition of 'genetic analysis' (*genetische Analyse*) (4,23 GTG) applies to analyses carried out in laboratories only raises the question whether the risk calculations,

which PG companies carry out in-house and which represent the main piece of information sold to consumers, could be seen as falling within the remit of the GTG;

(2) In light of the broad scope of traits and characteristics included in test batteries of PG-companies (including eye colour, photonic sneeze reflex, ear wax consistency) it is unclear whether PG-tests can be classified as 'genetic analysis on humans' (*genetische Analysen am Menschen*) which according to § 65,1 GTG are carried out 'for medical purposes' (*zu medizinischen Zwecken*).

With regard to the future development of the PG-market the report sketches three possible scenarios: a) an ongoing and increasing orientation of PG-companies towards collaborations with registered physicians and therefore a gradual erosion of the DTC-arm of the PG-market (this development has already become apparent in the US, in reaction to recent activities on the side of the Food and Drug Administration [FDA]); b) a stronger inclusion of genetic counselling into standard PG-testing services; c) a continuing increase of importance of partnerships between PG companies and clinical institutions, both for diagnostic, preventive, and research purposes. A disappearance of the PG-market is deemed an extremely unlikely scenario.

The author of this study suggests that attempts to prohibit DTC PG-tests would not be the most suitable and efficient way to respond to ethical and societal challenges raised by the PG-phenomenon. (In addition, the effectiveness of such prohibitions is dubious due to the transnational and virtual setting of the PG-market). Measures to raise public awareness and provide informational and educational material about PG-tests to the public, and to medical and paramedical professions, would be better suited to minimise the risk of unintended consequences of PG-testing. Last but not least, it is possible that the limited predictive value of these tests, which have been identified as a problem in the literature, could in contrast lead to the desirable outcome of giving people reason to have more sober and realistic expectations of what genetic information can actually say about individual future health. This would mean that PG-tests have the potential to contribute to a demystification of genetic data.