

Report on the Workshop *Legal regulation for genetic testing (WS10)*

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Milan Macek welcomes the participants of the workshop and concludes that this first workshop about legal aspects in the ESHG-Conference programme shows growing awareness that reflection on the legal side of scientific developments in the human genetics is needed. The objective of this workshop is to give an overview of EU-legislation and to highlight some legislative similarities and differences between European countries by giving examples of national legal provisions related to genetic testing (Austria, Germany, Portugal, Switzerland), as well as present recent initiatives of the Council of Europe. In addition, a legal case will be presented that shows how genetic knowledge may turn up (wrongly) in court.

Laurence Lwoff – EU

The objective of the Council of Europe regarding activities in bioethics is to find the necessary balance between scientific/medical progress and protection of the human being. The Oviedo Convention concerning Genetic Testing for Health Purposes (1994) gives the common framework for the protection of human rights regarding applications of biology and medicine. Recently the Additional Protocol concerning Genetic Testing for Health Purposes (the Protocol) is adopted (May 7th 2008). This Protocol is the first international legally binding instrument addressing this subject and was felt to be needed for various reasons: 1) the capacity of genetic analysis exceeds the capacity to act on it; 2) understanding the implications and results of tests is difficult and might obscure informed consent, and 3) entail possible risks for the privacy of persons and their families (e.g., discriminatory effects by employers/ insurance companies, breach of confidentiality and violation of the right (not) to know). The Protocol regards “tests involving analysis of biological samples, to identify genetic characteristics inherited or acquired during early prenatal development”. It does not apply to tests on embryos or fetuses or for research purposes. Central are the quality requirements for tests (analytical and clinical validity, but also clinical utility as key criteria), laboratories and professionals. The general rule is that genetic testing for health purposes may only be carried out under individualized medical supervision. The only exception to this general rule (e.g., “over-the-counter tests”) applies to tests that would not have serious implications for the person tested or for procreation choices; of course the requirements of nature and quality of prior information, free and informed consent and adequate genetic counselling apply here as well. Companies cannot get round this Protocol easily by asserting that their test offer is “not for health

purposes". The Protocol has already been implemented by some EU-member States, and serves as well as guideline for legislation for non-EU States and professionals' Codes of Conduct.¹

Dietmar Vybiral – Austria

The Austrian Gene Technology Act (1995) (the Act), which regulates molecular genetic testing on humans, was adopted in 1995. To adjust the law to current state of the art, an amendment on Gene Testing and Gene Therapy on humans (section IV) came into force in 2005. Based on the potential consequences, the Act distinguishes 4 types of genetic tests of which the first two serve to determine a present disease or to prepare for (the course of) a therapy, based i) on information about changes/variants in genes and DNA fragments or ii) on germline mutation; and the last two types determine a predisposition for a disease (genetically based and future onset) or carrier status, for which iii) therapy is possible, or iv) therapy is not possible. Only human/medical genetic specialists are allowed to perform tests iii) and iv), and a written and rather detailed informed consent is required for tests ii), iii) and iv). This classification needs regular updating and explanation to be sure that new technologies and tests which are intended to be covered by the Act, indeed are.

Quality requirements are set for pre- and post-test genetic counselling and laboratory management. Labs need approval (by the Minister of Health; currently 60 approved labs) to perform genetic tests, they are inspected by experts and can lose their approval if they do not meet the requirements. The use of collected genetic data is very restricted and employers and insurers are prohibited to ask for/use it. The Advisory Board on Biotechnology has published more detailed rules regarding genetic testing (e.g., for pharmacogenetic purposes) in the Austrian Book of Biotechnology (Gentechnikbuch)², which has the legal status of an objectified expert opinion and can as 'published ordinance' enter into force as a law.

Holger Tönnies – Germany

In January 2010, after a long discussion process, the Act on Human Genetic Testing (Gendiagnostikgesetz: GenDG)³ was adopted in Germany. This GenDG strives to regulate the use of human genetic diagnostics, which has recently changed both quantitatively and qualitatively: new genetic tests like next generation sequencing and arrays will be more available. Furthermore, the role of genetics in medicine will change: genetics will be applied for prediction, prognosis, prevention and mass screening for both monogenetic and multifactorial diseases. Pharmacogenetics is also a fast developing field of interest. Meanwhile, comprehensive genetic knowledge in the general public is lacking. To fulfill the state's duty to protect human dignity and to ensure individuals' right to self-determination, requirements for genetic examination and analysis are aimed at the prevention of disadvantages (e.g., discrimination) of genetic characteristics. As international basis for GenDG served, among other, the Oviedo Convention on Human Rights and Medicine and the Additional Protocol (see: Lwoff) and the International Declaration on Human Genetic Data (2003). GenDG-regulates partly even beyond these international standards. It proved to be hard to formulate right definitions that cover the intended area. GenDG applies to genetic testing of different subjects: persons, embryos and fetuses, and for different purposes: medical, descent, insurance and employment. Research purposes are not part of the GenDG.

¹ Further reading: Lwoff, L. Council of Europe adopts protocol on genetic testing for health purposes. *Eur J Hum Genet* 2009;17:1374-7. Text and Explanatory Memorandum Additional Protocol available at: <http://conventions.coe.int/Treaties/Html/2-3.htm>.

² Available at: www.gentechnik.gv.at (only in German).

³ Gendiagnostikgesetz Bundesgesetzblatt. Jahrgang 2009 Teil I Nr. 50, ausgegeben zu Bonn am 4. August 2009, 2529-2538; available at: http://www.bgbl.de/Xaver/start.xav?startbk=Bundesanzeiger_BGBI&bk=Bundesanzeiger_BGBI&start=//*/%5B@attr_id=%27bgbl109s2529.pdf%27%5D.

Genetic testing for criminal procedures and infection protection are regulated separately. §23 GenGD regulates the independent, interdisciplinary Genetic Diagnostic Commission (GEKO) to establish guidelines, in reference to 'generally accepted status of science and technology'. This GEKO and its working groups meet regularly to discuss new developments in genetic testing, and to establish new guidelines. These permanently updated guidelines can be said to be the living part of the law.

Jorge Sequeiros – Portugal

The Portuguese "Personal Genetic Information and Health Information Law" was published in January 2005, after five-years preparation and discussion. The law has a broad scope: it aims at protection of sensitive genetic information and sets requirements for genetic testing (in health care) and counselling, for genetic databases and biobanks, for biomedical research and for non-discrimination. Only when used for confirmation/exclusion of a clinical (postnatal, prenatal or preimplantation) diagnosis or for pharmacogenetics use, is genetic information considered to be "medical in nature".

For presymptomatic and susceptibility testing a much stricter regime applies. Carrier, presymptomatic and susceptibility testing must be preceded by counselling (proportionate to disease severity, age-at-onset and treatment) and requested by a medical geneticist.

Health information is defined to be the property of the person concerned and can only be used for aims mentioned in law, e.g., health care and health research. Genetic information (obtained by testing or other means) is explicitly not to be used by insurers, employers (except for health or safety purposes) or adoption agencies. General patients' records should only contain information of immediate interest for the patient's current health: other results from testing (e.g., in healthy subjects) must be kept in separate files at medical genetic services. Identifiable samples cannot be used for commercial purposes or stored by commercial entities. If coded samples are necessary, a public institution should keep the identifying codes. Biobanks must have a health-care or health-research purpose only and have to involve a medical geneticist whenever communication of results (re-contacting) is foreseeable.

The government is planning to regulate the offer of genetic testing by public and commercial laboratories, including outside the context of medical services and genetic counselling, as in direct-to-consumer testing.

Michael Morris – Switzerland

The Swiss federal law on Human Genetic Testing (2004)⁴ came into force April 1st 2008. This law was initiated by "Popular Vote" in order to protect human dignity and personality by preventing improper genetic testing and improper use of genetic data. Therefore, this law regulates the quality of performing genetic tests and interpreting their results, and explicitly bans discrimination on grounds of one's genetic material. It stipulates different conditions of genetic testing in various contexts: medical care, employment, insurance, and liability. Directives for direct-to-consumer testing are set by the Expertenkommission für genetische Untersuchungen beim Menschen (GUMEK).⁵

Requirements in the law for diagnostic testing are less stringent than for prenatal or presymptomatic genetic testing with regard to prescription (by medical doctors vs. doctors with appropriate postgraduate training), informed consent (verbal vs. written) and genetic counselling (adequate information vs. detailed prescription of topics and obligatory "time for reflexion" between counselling and actual testing). Compliance with these requirements is however hard to check. An annex to the Swiss Law on Health

⁴ DE:GUMG; FR: LAGH; IT: LEGU => http://www.admin.ch/ch/d/sr/c810_12.html. ENG (no legal force) => Google: federal LHGT.

⁵ <http://www.bag.admin.ch/themen/medizin/00683/02724/04638/index.html?lang=de>.

Insurance (LAMal) contains a positive and limitative, albeit rather unsubstantial and discriminatory, list with diagnostic tests which are to be reimbursed by health insurance. Quality Assessment and accreditation for laboratories performing genetic tests is encouraged, but not formally required. Thus, the law enables to practice medical genetics and provides for quality requirements, but shows hiatus as well.

Francesca Forzano – a legal case

A controversial legal proceeding in Italy went on as follows. Mr F was murdered by Mr A, who was affected by schizophrenia and actively psychotic at the time of the crime, for he had discontinued his psychotropic medication. He was judged (on first level) guilty of murder, but the sentence (9 years) was reduced because of his mental illness. In appeal court, a new expert assessment also including tests for various genetic variants associated with antisocial behaviour was requested. Experts' opinion was, inter alia, that Mr A carried at least one allele associated with a significant increase of the risk of aggressive and impulsive behaviour. Taking into account this fact, the appeal court further reduced the sentence from 9 to 8 years, because Mr A was considered even more vulnerable under stressful conditions than previously reckoned. This sentence was heavily criticized, because 1) an individual should be judged on the basis of evidence-based information and not on the basis of probability interpretation; 2) the possibility to use genetic variants for evaluating one's actual mental capacity at a specific time is not established; 3) the genetic variants do not add to the evaluation of the phenotype itself; and 4) testing for most of these variants is not properly evaluated in terms of clinical validity and utility, while valid and reliable evidence and a robust test for the genetic factor concerned is essential. To prevent confusion about the term "responsibility" by legists and geneticists, it might be preferred to avoid using this term by the latter at all: "a gene never exerts any effect in terms of 'responsibility', and this word from a philosophical and legal register should not be used to describe biological mechanisms."⁶ Anyhow, a connection between legal responsibility and genes being "responsible" for some behavioural effect is improbable to make and should be dealt with very cautiously.

Final remarks

There is a generally felt need for separate regulation of genetic testing. Rather than demonstrating 'genetic exceptionalism', this recognizes that the infrastructure and the possibilities of (using and abusing information generated by) genetic testing justify specific regulation. National legislation of genetic testing shows similarities with regard to its aim and domains for which quality requirements are provided. The manner and degree of specification differs. Some choose to establish only general rules in the law and leave the details to lower levels of regulation or to guidelines made by, e.g., specialist committees in order to enable flexible issuing of rules adapted to new developments. Others offer very detailed definitions and rules in the law itself, which makes legislation seemingly more enduring, but yet also less suitable for the dynamic practice. Although all legislation acknowledges that good quality in every phase and part of genetic testing is essential (e.g., pre- and post-test genetic counselling, informed consent procedures, laboratory requirements), this is enforced differently: required vs. encouraged accreditation of laboratories, systematic vs. occasional inspection by authorities. The ESHG recognizes the importance of (studying) the legal side of scientific developments in human genetics and wants to support this by, e.g., making easily available EU and national legislation on genetic testing as a frame of reference.

⁶ Forzano F, Borry P, Cambon-Thomsen A, Hodgson SV, Tibben A, de Vries P, van El C, Cornel M. Italian appeal court: a genetic predisposition to commit murder? *Eur J Hum Genet* 2010;18(5):519-21.