Editorial

Genetic Testing for Health Care Purposes, a Council of Europe Protocol

1. Introduction

The understanding of the human genome, the function of human genes, their way of interacting, also in relation to other factors (microbes, environment and lifestyle), has brought opportunities for the prevention and treatment of disease. Non-somatic genetic testing and screening uncover information on hereditary or congenital genetic characteristics. The testing technology may be applied to a living person, to an embryo and foetus (pre-implantation and prenatal diagnosis) and after death. The purpose of genetic testing is to diagnose a disease (including the confirmation of a suspected disease), to predict the possibility of future illness (pre-symptomatic and susceptibility testing), to detect carrier-status of a genetic defect, and to predict responses to therapy. Simultaneously with the development of genetic technology (international) genetic registries have been set up for the purpose of individual care, research and statistics. Collections of human biological samples (and associated data) have become essential for research, development and application of genetic testing technology.

Genetic testing has become an integrated part of health care. It is known to be applied for non-health purposes as well. They include employment, insurance and identification purposes.

Genetic testing on individuals and in the context of screening is a rapidly growing sector. Advances in science will provide more and more information on relevant factors in relation to susceptibility for disease development, thus eventually allowing for individualised prevention and treatment. As with other medical examinations, different genetic tests may reveal different kinds of information,

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1) Somatic genetic mutations take place in non-germinal cells and are not transmissible to offspring. They are not dealt with.

2) Screening involves a systematic offer to a seemingly healthy (target group) of the population for the purpose of early identification of health risks (latent disease or risk factor for future occurrence of a disease) in order to facilitate prevention and early treatment. A positive outcome of a screening test requires confirmation.

3) Employment related testing may have a health purpose as well.
ranging from low to high information content, that brings accordingly low and high informational risk, depending on whether heredity is a contributing factor or plays a central role.

Market structures are expected to change according to demand and an increase of private (commercial) laboratories and companies offering genetic testing, easy-to-use DNA kits and self-test kits is expected.\(^4\)

Obviously, as with health services in general, this field of medicine also necessitates an assurance of the validity and utility of genetic tests, strict adherence to quality standards by the laboratories performing the testing, and skilled and knowledgeable health personnel involved with genetic testing and counselling, also in relevant ethics and law. It is the role of governments to ensure that genetic services function at consistently high standard.

2. Ethical and Legal Concerns

The advances in genetic testing technology have fuelled debate on its effects on the human rights of individuals. Individual rights and interests of relatives easily conflict. Individual informational privacy is difficult to maintain where interests of relatives are involved, as is the case with genetic information. Interests of relatives put the autonomy of the individual under pressure. Genetic testing technology is prone to misuse and abuse. These and other risk factors for individual human rights have given rise to questions and dilemma’s. For the individual in relation to private life (the right to know vs. the right not to know, confidentiality vs. duty to share information); for the professionals in relation to professional standards (medical secrecy vs. the duty to warn). Confidentiality, the right not to know and voluntariness are easily compromised when genetic technology and genetic information is used for non-medical purposes (employment, insurance, identification). Insufficient clarity about the legal position of the source of stored human tissue over the use of his material for genetic purposes may decrease trust in health care professionals. Concerns have also been raised in relation to the use of genetic technology in the context of a screening programme. (Genetic) screening ought to be offered only under strict conditions. They include the condition that the screening programme should serve the (direct) interests of the participants.\(^5\) The availability of adequate treatment or other intervention is an essential criterion for selecting diseases suitable for screening.\(^6\)


\(^6\) Adequate is determined both by proven medical effect and ethical and legal acceptability. Council of Europe Recommendation R. 94 (11) on screening as a tool of preventive medicine.