



## **Explanatory Report to the Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes \***

Strasbourg, 27.XII.2008

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I. This Explanatory Report to the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes was drawn up under the responsibility of the Secretary General of the Council of Europe.

II. The Explanatory Report takes into account the discussions held in the Steering Committee on Bioethics (CDBI) and its Working Party entrusted with the drafting of the Protocol; it also takes into account the remarks and proposals made by delegations. The Committee of Ministers has authorised its publication on 7 May 2008.

The Explanatory Report is not an authoritative interpretation of the Protocol. Nevertheless, it covers the main issues of the preparatory work and provides information to clarify the object and purposes of the Protocol and to better understand the scope of its provisions.

### **Introduction**

1. Remarkable progress has been made in the field of human health thanks to research in biology and medicine. In that respect, genetics is one of the sciences seen as most promising. Knowledge on the human genome has been the source of considerable advances, in particular the development of genetic tests involving analysis enabling the identification of genetic characteristics responsible for a disease (monogenic diseases), or involved in its development (multifactorial diseases the development of which is also influenced by other factors). These tests make it possible to diagnose or to confirm the diagnosis in a person already presenting symptoms. But they also make possible the identification of genetic mutations responsible for a disease which only develops later in life, or of a predisposition to a disease before symptoms appear.

2. Early identification of genetic characteristics by a test can present a health benefit, if it makes it possible to take preventive measures or to limit the risks by modifying the behaviour, life style or environment of the person concerned. However, for most genetic diseases, such possibilities are still very limited. Furthermore, the results of genetic analysis are often complex and a proper understanding of their implications is, in many cases, difficult to understand for the persons concerned.

3. Finally, genetic tests are to become more and more an integral part of medical practice, but at the same time a direct commercial offer for genetic tests outside any health system is developing.

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(\*) The Treaty of Lisbon amending the Treaty on European Union and the Treaty establishing the European Community entered into force on 1 December 2009. As a consequence, as from that date, any reference to the European Community shall be read as the European Union.

4. This Additional Protocol to the Convention on Human Rights and Biomedicine concerning genetic testing for health purposes builds on the principles embodied in the Convention with a view to ensuring protection of people in the specific field of genetic testing for health purposes.

5. The purpose of the Protocol is to define and safeguard fundamental rights of the persons concerned by genetic testing for health purposes.

### **Drafting of the Protocol**

6. In 1991, in its Recommendation 1160, the Council of Europe Parliamentary Assembly recommended that the Committee of Ministers “envisage a framework convention comprising a main text with general principles and additional protocols on specific aspects.”

7. The Convention on Human Rights and Biomedicine was adopted by the Committee of Ministers on 19 November 1996. The same year, the Committee of Ministers instructed the Steering Committee on Bioethics (CDBI) “to draw up a Protocol to the Convention on Human Rights and Biomedicine concerning the problems relating to human genetics...” and accordingly invited the CDBI “...to start work on it as soon as possible, taking also into account questions relating to the use and protection of the results of predictive genetic tests for purposes other than health or scientific research linked to health.”

8. At its 12th meeting (17–19 June 1997), the CDBI appointed Dr Stefan WINTER (Germany), Chairman of the Working Party on Human Genetics (CDBI-CO-GT4) and decided to hold a first exploratory meeting on 12–14 November 1997. In addition to Dr Winter, were also appointed to take part in the exploratory meeting, Dr Elaine GADD (United Kingdom), Mrs Ruth REUSSER (Switzerland) and Mrs Lena JONSSON (Sweden). A workshop on “Medical, ethical and social aspects of new and perspective developments in genetic research” was then held on 21 – 22 April 1998 in Leuven (Belgium), as well as a hearing of European patient organisations on 8 June 1998 in Strasbourg. On the basis in particular of the conclusions of those different meetings to start its work, the Working Party held its first meeting in September 1998.

9. When this work was undertaken, the CDBI was also working to the finalisation of the Additional Protocol to the Convention on Human Rights and Biomedicine concerning Transplantation of Organs and Tissues of Human Origin (opened for signature on 24 January 2002) and the Additional Protocol to the Convention of Human Rights and Biomedicine concerning Biomedical Research (opened for signature on 25 January 2005).

10. In February 2003, a working document presenting the outcome of the discussions carried out so far by the Working Party on Human Genetics (CDBI-CO-GT4) on applications of genetics for health purposes with a view to the elaboration of the Additional Protocol, was made public for consultation purposes, under the responsibility of the Working Party (CDBI-CO-GT4).

11. In December 2003, the CDBI revised the terms of reference of the CDBI-CO-GT4 and its composition, taking into account the need for different expertises within the Working Party to elaborate provisions concerning the fields of employment and insurance.

12. At its 27th meeting (18–22 October 2004), the CDBI agreed to focus the Protocol on genetic testing, considering that gene therapy was essentially a research concern and that the value added by the provisions that could be put in the Protocol remained extremely limited. At the same meeting, the CDBI agreed to exclude from the scope of the Protocol, genetic testing for identification purposes. A similar decision was also taken concerning research, already covered in general by the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research, adopted by the Committee of Ministers on 30 June 2004.

13. At its 29th meeting (17–21 October 2005), considering the importance of the ethical questions raised by prenatal genetic testing, but also their specific and complex nature, the CDBI confirmed its decision taken in 2001 to exclude genetic testing on the human embryo and foetus from the scope of the Protocol and considered addressing them independently.

14. The CDBI decided, at its 30th meeting (2–5 May 2006), to split the Protocol and to bring out separate instruments dealing with genetic testing for health purposes and genetic testing for employment and insurance purposes.

15. The Protocol was approved by the CDBI at its 32nd meeting (4–8 June 2007) under the Chairmanship of Professor Elmar DOPPELFELD (Germany). The Parliamentary Assembly gave the opinion No. 267 (2008) on the Protocol, on 24 January 2008, on the basis of a report elaborated by Mr. Wodarg for the Committee on Culture, Science and Education, and of an opinion prepared by M. Haibach for the Committee on Legal Affairs and Human Rights. The Protocol was adopted by the Committee of Ministers on 7 May 2008.

## **Commentary on the articles of the Protocol**

### **Title**

16. The title identifies this instrument as the “Additional Protocol to the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Applications of Biology and Medicine (Convention on Human Rights and Biomedicine), concerning Genetic Testing for Health Purposes.”

### **Preamble**

17. Protection and guarantees in the fields of biology and medicine, including human genetics, are provided by the Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine (Convention on Human Rights and Biomedicine) hereafter the “Convention”.

18. After the Protocol on the prohibition of cloning human beings, the Protocol concerning transplantation of organs and tissues of human origin, and the Protocol concerning biomedical research, the Additional Protocol on genetic testing for health purposes supplements further the provisions of the Convention. The Protocols are designed to address ethical and legal issues raised by present or future scientific advances through the further development, in specific fields such as genetic testing, of the principles of the Convention.

19. The Preamble of this Protocol reaffirms the aims of the Council of Europe, and of the Convention. It refers to the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data of 28 January 1981. It also recalls the work carried out by other intergovernmental organisations, and in particular the Universal Declaration on the Human Genome and Human Rights, endorsed by the General Assembly of the United Nations on 9 December 1998.

20. It recalls the particular bond existing between the members of the same family, which is due to the genetic characteristics that they share.

21. It underlines the role of progress in biomedical sciences in reducing morbidity and mortality and in improving the quality of life, and notes the potential benefit of genetics in the field of health. However, it also acknowledges the concerns raised regarding possible improper uses of the information generated by genetic testing.

22. Furthermore, it takes in due consideration the previous work of the Committee of Ministers and the Parliamentary Assembly concerning genetic services, which was taken into account in the elaboration of this Additional Protocol.

23. The preamble reaffirms the commitment of the Parties to take, with regard to genetic testing for health purposes, all necessary measures to safeguard human dignity and the fundamental rights and freedoms of the person. It highlights some of the fundamental principles that underline this commitment:

- the prohibition of any discrimination, in particular based on genetic characteristics;
- equitable access to genetic services of appropriate quality.

## **CHAPTER I – Object and scope**

### **Article 1 – Object and purpose**

24. This article is based on the wording of Article 1 of the Convention on Human Rights and Biomedicine. It specifies that the object of the Protocol is to protect the dignity and identity of all human beings, and guarantee everyone, without discrimination, respect for their integrity and other rights and fundamental freedoms with regard to such tests as specified in this Protocol.

### **Article 2 – Scope**

25. This article specifies the scope of the Protocol and defines the main terms it employs.

26. Paragraph 1 states that the Protocol applies to tests, carried out for health purposes, which involve analysis of biological samples of human origin, and specifically aim to identify genetic characteristics of a person which are inherited or acquired during early prenatal development.

27. Therefore, not covered by the Protocol are the genetic tests carried out for identification purposes, such as those carried out within the framework of a medico-legal expertise or in view of establishing parentage, except if this research is carried out for medical purposes.

28. Furthermore, the requirement that the test involves the analysis of a biological sample excludes as such the collection of genetic information through family history.

29. The notion of “genetic test” is based here on two elements: the method used and the purpose of the test. It is to be understood as a procedure including removal of biological material of human origin, where relevant, as well as the analysis of the personal information obtained there from. This procedure aims specifically to identify genetic characteristics of a person which are inherited or acquired during early prenatal development. These genetic characteristics cover those already present in the gametes of the parents and therefore transmitted by the latter, as well as those which appear during the early stage of prenatal development before the differentiation of the germ line. It is sometimes referred to the genetic characteristics inherited or acquired during early prenatal development as “genetic characteristics transmissible to descendants”. The genetic modifications acquired during lifetime by only certain somatic cells due for example to external factors in the environment, are therefore not covered.

30. The Protocol covers any genetic test carried out for health purposes on a person whether living or dead (in the interests of the latter’s family members), or on biological material of human origin. This includes diagnostic, predictive or healthy carrier tests as well as pharmacogenetic tests. Genetic tests offered in the framework of a genetic screening programme are also covered by the Protocol.

31. The Protocol excludes from its scope genetic tests on the human embryo and foetus. Therefore, preimplantation (PGD) and prenatal genetic diagnosis (PND) are not covered, including tests on polar bodies (small haploid cells containing a single set of chromosomes – produced by the ovocyte during meiosis – the process whereby reproductive cells divide to produce gametes), as well as tests on components of embryonic or foetal origin (such as DNA or cells) present in the mother's blood to obtain information about the foetus or embryo.

32. Genetic tests carried out for research purposes are also excluded from the scope of the Protocol. It should, however, be noted that Article 12 of the Convention also applies to predictive tests for health research purposes, and states that such tests may only be carried out subject to appropriate genetic counselling.

33. It has to be noted that some genetic tests, even at an experimental stage, may reveal personal information relevant to the health of the person concerned. The Additional Protocol to the Convention on Human Rights and Biomedicine, concerning biomedical research, provides in Article 13, paragraph 2.v. on information for research participants that these persons must be given specific information on arrangements for access in particular to "information relevant to the participant arising from the research". Furthermore, the Appendix to that same Protocol provides in paragraph xv that the ethics committee must be informed of the "arrangements foreseen for information which may be generated and be relevant to the present or future health of those persons who would participate in research and their family members". This applies precisely to genetic tests. As the Explanatory Report comments under this provision (paragraph 57), "... Because proper counselling and other health care assistance may be necessary to explain the nature of the results and the options available to the participant, ...". It is therefore good practice to provide this information and, where appropriate, to offer genetic counselling, according to the principles set out in Chapter IV of this Protocol.

34. More often than not the analysis of biological samples are analysis of chromosomes, or analysis concerning DNA or RNA. However, tests using analysis of any other element enabling information to be obtained which is equivalent to that obtained by the methods referred to above for the determination of genetic characteristics which are inherited or acquired during early prenatal development are also considered to be covered by the Protocol.

35. The term "equivalent information" must be understood as referring to information directly linked to the genetic characteristics sought. The analyses in question enable direct information to be obtained on the genetic heritage of the person on whom the test is carried out. This is the case in particular with analysis of gene expression products such as proteins. A distinction must be drawn between analysis providing information of that kind and analysis simply providing indications on genetic characteristics without enabling a direct link with them to be established. These indications alone do not provide a sufficient basis for confirming or otherwise the presence of a genetic modification, but they may call for further investigations. This distinction may be illustrated, for example, by the case of a genetic modification leading to hypercholesterolemia in which the gene involved is the MTP (Microsomal Triglyceride Transfer Protein) gene and its expression product: the MTP protein. The action of this protein results in a change in the cholesterol level in the blood. However, blood analysis of the cholesterol level will only give an indication on the genetic characteristics of the person concerned, since the cause of the hypercholesterolemia may be unconnected with the MTP gene and be due to another factor. Consequently, this analysis does not provide "information equivalent" to that provided by DNA, RNA or even the MTP protein, the MTP gene's expression product. Generally speaking, the definition of the types of analysis specified in paragraph 3a. does not include analysis of elements that are not directly linked to the genetic characteristics sought.

36. "Biological sample" refers to biological material removed for the purpose of the genetic test considered, but also any biological material originally removed for another purpose, on which the test is performed.

## **CHAPTER II – General provisions**

### **Article 3 – Primacy of the human being**

37. This article affirms the primacy of the human being concerned by genetic tests covered by the Protocol, over the sole interest of society or science. Priority is given to the former, which must in principle take precedence over the latter in the event of conflict between them.

38. The whole additional Protocol, the aim of which is to protect human rights and dignity, is inspired by the principle of the primacy of the human being, and all its articles must be interpreted in this light.

### **Article 4 - Non-discrimination and non-stigmatisation**

39. Paragraph 1 repeats the wording of Article 11 of the Convention on Human Rights and Biomedicine. Non-discrimination is an individual right enshrined in Article 14 of the Convention for the Protection of Human Rights and Fundamental Freedoms. Under Article 14 of this Convention, the enjoyment of the rights and freedoms set forth in the Convention must be secured without discrimination on any ground such as sex, race, colour, language, religion, political or other opinion, national or social origin, association with a national minority, property, birth or other status. The provisions of Article 11 of the Convention on Human Rights and Biomedicine, repeated in Article 4, paragraph 1 of this Protocol, add the person's genetic heritage to this list.

40. The concept of discrimination therefore relates to a difference in the treatment of the person concerned. Yet not all differences in treatment necessarily amount to discrimination. In particular, positive measures that may be implemented with the aim of re-establishing a certain balance in favour of persons at a disadvantage because of their genetic heritage are not regarded as discrimination. The criteria for assessment, according to the case-law of the European Court of Human Rights on Article 14 of the Convention, are the relevance and legitimacy of the aim pursued and the reasonable relationship of proportionality between that aim and the means used.

41. The Article also requires, in its paragraph 2, that appropriate measures be taken to prevent stigmatisation of individuals or groups in relation to genetic characteristics.

42. A distinction can be drawn between stigmatisation and discrimination, in that stigmatisation is not necessarily relevant to the exercise of an individual right. The concept of "stigmatisation" rather relates to the way in which a person or group is perceived on the basis, in this case, of their genetic characteristics, whether these exist or are thought to exist. It takes, in particular, the form of words or acts that negatively label a person or group of persons on account of their known or supposed characteristics.

43. Possible measures to prevent stigmatisation include general information campaigns on the human genome and its characteristics and on advances in our knowledge of human genetics, aimed at the general public as well as incorporated into education and training curricula. Parties should encourage such initiatives.

44. The provision of more targeted and more specific information can also be included by the competent authorities concerned in the setting up of a screening programme. Problems of stigmatisation may indeed arise with regard to those taking part in such a screening programme, or other members of the population or sub-group offered the screening. Screening programmes of this type are aimed at detecting or excluding, by means of a genetic test, the presence of certain genetic characteristics linked to a disease. The perception of the disease in question and the interpretation that could be made of the purpose of the screening could result in the individuals concerned being stigmatised. Particular attention should therefore be paid to the information and communication aspects of such programmes in order to limit the risk of these individuals being stigmatised.

## CHAPTER III – Genetic services

### Article 5 - Quality of genetic services

45. This article defines an aim and imposes an obligation on States to take measures to achieve it. The aim is to ensure an appropriate quality of genetic services. The purpose of these services, be they public or private, is to respond to the needs of individuals and families wishing to know whether they are at risk of developing or transmitting a disease or disorder with a genetic component, or who are faced with such a disease or disorder. This includes in particular providing information and, where appropriate, genetic counselling, carrying out genetic tests and interpreting their results, ensuring care for the persons concerned and their families, including preventive care, as well as training the persons involved in providing genetic services.

46. This article defines specific requirements at three different levels: the genetic test, the laboratory and the persons providing genetic services.

47. Paragraph a. requires Parties to take measures to ensure that genetic tests meet generally accepted criteria of scientific validity and clinical validity.

48. “Scientific validity”, also called “analytical validity”, refers to the way in which the test measures the characteristic it is designed to identify. In particular, this concept includes the capacity that the test will be positive if the genetic characteristic is present (analytical sensitivity), and negative if it is absent (analytical specificity).

49. The notion of “clinical validity” of a test is to be understood as corresponding to a measurement of the accuracy with which the test identifies or predicts a clinical condition. It is defined in terms of clinical specificity, sensitivity and predictive value.

50. “Generally accepted criteria” means those criteria which are widely recognised at international level.

51. According to this provision, it is the responsibility of the State to ensure the existence of a system (e.g. an approval system), which guarantees the reliability of a genetic test in respect of a determined disease, i.e. that its results with regard to the identification of particular genetic characteristics related to this disease are accurate and can be reproduced.

52. The “quality assurance programme” referred to in paragraph b concerns general quality controls on laboratory procedures rather than specific genetic tests. This requirement also applies to procedures undertaken in the framework of screening programmes. An accreditation system constitutes, for example, a generally effective measure to satisfy quality assurance requirements in particular for laboratories carrying out complex cytogenetic or molecular genetic analyses. By accreditation is meant, a procedure formally establishing the competence of the laboratory to carry out genetic tests.

53. Laboratories shall be monitored regularly, preferably by means of an external structure, to make sure they are complying with the established rules. This monitoring aims to ensure in particular respect of the confidentiality of data and security of the biological samples, the quality of the procedures and the specific scientific and technical skills of the staff involved.

54. In this context, it can be useful to refer to the guidelines elaborated in this field by intergovernmental bodies.

55. In paragraph c, the term “persons providing genetic services” includes medical doctors, nurses and other health care professionals as well as non-medical staff such as biologists and technicians working in the analysis laboratories. It is required that they have “appropriate qualifications”, which shall be understood as taking into account possible qualification systems in place at national level and including in-service training. As genetic tests are destined to become an increasingly regular part of ordinary medical practice, medical genetics, including what concerns counselling, should in particular be part of the education and training programme of all health care professionals.

#### **Article 6 - Clinical utility**

56. This article emphasises the importance of taking into account the clinical utility of a genetic test as an essential criterion for deciding to offer this test to a person or a group of persons.

57. "Clinical utility" is to be understood by the value of the test results in guiding the person concerned in his or her choices regarding prevention or therapeutic strategies. It is therefore a particularly important factor in deciding whether or not it is appropriate to offer a genetic test.

58. Criteria generally considered for determining the clinical utility of a test may be grouped in two large categories:

– Criteria concerning the test:

– the “service rendered” by the latter (in particular the value of the test results to determine the possible medical measures in terms of prevention or treatment);

– the circumstances in which the test is offered (quality and accessibility of the genetic services available, including genetic counselling, etc);

– Criteria relating to the situation of the individual to whom the test is offered.

59. The measure of the clinical utility of a test may vary from one individual to another, depending on that individual’s situation, with social and cultural aspects also being taken into account. For example, awareness of relevant information for the health of his or her family members, or information on the risk of developing the disease, even in the absence of a prevention or treatment strategy, could, for a particular individual, be beneficial, including in terms of well-being; this will play a part in determining the clinical utility of the test in question for that individual.

60. Taking account of the clinical utility of a test can be regarded as an integral part of good medical practice with regard to any decision to carry out the test under individualised medical supervision.

61. The provision of this article is of special importance for tests proposed without such medical supervision, and for the planning of screening programmes.

62. Existing evidence on a genetic test’s clinical utility have thus to be available, in particular, to the health professionals and the persons concerned by that test. Such evidence should be obtainable from the laboratories carrying out the test.

#### **Article 7 – Individualised supervision**

63. Paragraph 1 establishes the general rule according to which a genetic test may only be carried out under individualised medical supervision. The concept of “medical supervision” shall be understood as referring to a process within which the genetic test will take place.



64. This provision is driven by the concern, in particular, to enable the person concerned to have suitable preliminary information with a view to an informed decision regarding the carrying out of this test and, if appropriate, to have access to an appropriate genetic counselling. A precise evaluation of the situation of the person concerned, involving direct contact with him or her, is a determining element in that respect. A mere telephone conversation with a medical doctor, for example, does not allow for such an evaluation.

65. The conduct of a genetic test for health purposes must be in response to a specific request made on the basis of a precise evaluation of the situation of the person concerned, carried out by a medical doctor.

66. Paragraph 2 provides for exceptions to the general rule laid down in paragraph 1, on condition that appropriate measures, taking into account the way the test will be carried out, are provided to give effect to the other provisions of this Protocol. The main purpose is to ensure, in particular, compliance with the provisions concerning the nature and quality of the prior information, free and informed consent and genetic counselling.

67. The exceptions under consideration do not concern the performance of a test on a particular individual but rather readily identifiable test device for which the genetic characteristics it is meant to identify would be specified. The genetic tests concerned may be carried out by a laboratory after the biological material has been removed by a professional or by the person concerned him or herself who then sends it to the laboratory. They may also be tests entirely carried out by the person concerned with a kit enabling him or her to remove the biological sample as well as to carry out the analysis.

68. The objective is the protection of the person concerned. It is left to each State to determine how to implement this provision effectively. The same applies to the procedure to be followed and to the authorities or bodies involved in the decision to authorise a test complying with the legal requirements for marketing it, to be carried out without individualised medical supervision. Particular attention must be paid in this process to the importance of the potential implications of the test considered for the persons on whom it would be carried out or for the members of their family, the ease of interpretation of the results and, if appropriate, the treatment possibilities for the disease or disorder concerned. The envisaged measures to give effect to the provisions of this Protocol could be different depending on whether the test considered is fully carried out by the person concerned by means of a kit or whether the analysis is carried out by a laboratory.

69. The performance without individualised medical supervision of genetic tests on persons not able to consent, raises special concerns. States should bear these in mind when authorising, or not, direct access to such tests.

70. The correct interpretation of results and the guarantee of appropriate genetic counselling to understand their implications remain the main concern. It is considered, in this respect, that these requirements envisaged by the present Protocol cannot be satisfied outside of individualised medical supervision in the case of genetic tests with important implications for the health of the person concerned or of members of his or her family, or for choices concerning procreation. The test results may be particularly complex to interpret and require, for example, additional medical information or information concerning family history to be taken into account. With many predictive tests, even if they reveal a strong likelihood of developing a particularly serious disease, the time of onset, if at all, and the severity of the symptoms will often remain uncertain. Finally, understanding the nature and implications of the test, including for family members, the possible psychological impact of the results on the person concerned and the often important decisions he or she is faced with requires such tests to be performed under individualised medical supervision.

## **CHAPTER IV – Information, genetic counselling and consent**

### **Article 8 – Information and genetic counselling**

71. In accordance with Article 5 of the Convention on Human Rights and Biomedicine, Article 8 of this Protocol stipulates in its paragraph 1 that any person on whom a genetic test is envisaged shall be provided with prior appropriate information. This preliminary information shall include in particular the purpose and nature of the test considered as well as the implications of its results for the person concerned. Where appropriate, especially in the case of a predictive test, information on the implications for members of his or her family must also be included. These requirements relate to all the genetic tests covered by this Protocol, including those covered by the provisions of Article 7 paragraph 2, envisaged outside of the individualised medical supervision.

72. The "implications" of the test results are to be understood, in particular, as covering the benefits and the risks, including at psychological level. Possible preventive or therapeutic measures and the constraints they entailed shall also be specified. The person shall also be informed of the consequences of not carrying out the test and, where appropriate, of the possible alternatives. To health consequences shall also be added, where applicable, those concerning future procreation choices, both for the person concerned and possibly for the members of his or her family. The possibilities open to the person shall also be explained to him or her, in particular, in accordance with the provisions of Article 16 of this Protocol, his or her right not to be informed of the results of the test. Also, the information must include the forms of support available to the person, in particular genetic counselling.

73. Those elements listed above are not meant to be exhaustive; it merely explains the essential tenor of the information to be provided. However, this information must be adapted to the test envisaged, as well as to the person concerned. The implications of test results for a person already showing symptoms of a disease or disorder (diagnostic test), for example, will be different from those of a predictive genetic test which will provide information about the person's future health. Furthermore, the person's individual characteristics – such as his or her age – must also be taken into account, for example when considering the possible consequences in terms of procreation choices. Moreover, at the request of the person concerned, additional information, clarification or precision must be provided in order to enable an informed decision.

74. The content of the information but also the form in which it is transmitted are paramount in guaranteeing a free and informed decision by the person concerned. The information must be sufficiently clear and comprehensible for the person, bearing in mind his or her level of knowledge, his or her education and psychological condition. Furthermore, like all information prior to a medical act, it must be conveyed in a fair and neutral manner, to enable the person concerned to reach a free and informed decision.

75. For genetic tests referred to in paragraph 2 of this article, oral communication of information is essential. In particular it makes it easier to facilitate and ensure proper understanding by the person concerned by answering his or her possible questions and supplying clarifications if necessary.

76. Information could also be provided concerning any possible subsequent use of the biological material removed which is foreseen. It would be preferable for this information not to be disclosed at the same time as that concerning the proposed test, in order to avoid confusion. It should be made clear that any subsequent use would require further consent distinct from that required for the genetic test.

77. Paragraph 2 refers to tests covered by Article 12 of the Convention on Human Rights and Biomedicine. These are tests predictive of a monogenic genetic disease or serving to detect a genetic predisposition or susceptibility to a disease. They also include tests which make it possible to identify the person tested as the healthy carrier of a gene responsible for a disease. That gene's presence will not affect the health of the person concerned, but may have implications for the health of his or her descendants.

78. The tests referred to here all relate to diseases, and do not therefore include pharmacogenetic tests.

79. The predictive nature of the information obtained with tests covered, the emotional impact for the person concerned of knowing about a genetic risk, the possible implications for family members and the often important decisions with which the person concerned may be faced, including where appropriate in relation to procreation choices, explain the importance of an appropriate genetic counselling for those tests.

80. Some diagnostic tests may also have important implications for decisions on procreation choices. Indeed they may provide information about the origin, possibly genetic, of the disease, the symptoms of which are observed in the person tested, which may, where appropriate, affect his or her descendants. For such tests, it is good practice that the person concerned also has access to appropriate genetic counselling.

81. The notion of "genetic counselling" is to be understood here as a communication and support process aiming to enable individuals and, where appropriate, families to make informed choices with regard to a genetic test and its implications.

82. It includes the provision of information prior to consent as required in paragraph 1. It also includes an offer of support before and, if appropriate, after the test, to the person concerned, to help him or her to deal with the implications of the test and its results, including, where appropriate, communication to family members of information relevant to their health, or procreation choices.

83. Genetic counselling is an individualised process taking into account, in particular, the psychological and family context of the person concerned and involving an exchange between him or her and the person providing the counselling. This support process may therefore vary in form and extent depending on the test considered but also on the particular significance of the information that the test is likely to provide for the person concerned or for members of his or her family. In certain cases, the person concerned would also benefit from psychological support provided by persons with appropriate competencies.

84. When the results of a test carried out on a person may have important health implications for members of his or her family, one must refer to Article 18 (Information relevant to family members) of this Protocol and to paragraphs 138 to 141 of this Explanatory Report.

85. At all events, genetic counselling shall not be delivered in a directive manner. However it can be difficult for the person providing the genetic counselling to avoid influencing the subject to some extent, because, among other things, of the counsellor's status, knowledge and experience and the subject's perception of them. At all events the person providing the genetic counselling must explain the different options open to the subject in such a way as not to influence unduly the subject's decision, which must be freely taken.

#### **Article 9 - Consent**

86. Paragraph 1 follows the wording of Article 5 of the Convention on Human Rights and Biomedicine, in requiring the free and informed consent of the person concerned before any genetic test is carried out. The test may be performed on a biological sample removed for that purpose, or on biological material originally taken from the person concerned for another purpose.

87. Because of the predictive nature of the results of the tests referred to in paragraph 2 of Article 8 and their potential implications for the person concerned and/or for the members of his or her family, the article provides for the consent to be documented. This is generally achieved through a signature by the person concerned. However, this does not necessarily rule out the possibility of using other forms of registration of the expression of consent as long as it attests the authenticity of the consent and permits to keep a permanent record of it. It should also be noted that if the elements of capacity to consent, appropriate prior information and voluntariness have not been satisfied, a signature on a form will not make the consent valid.

88. Freedom of consent implies that consent may be withdrawn at any time. The person must be informed of the possible consequences of withdrawing the consent. His or her decision must be respected. There is no requirement for withdrawal of consent to be made in writing or in any other specific form.

## **CHAPTER V – Persons not able to consent**

### **Article 10 - Protection of persons not able to consent**

89. The article establishes the rule that, subject to Article 13 of this Protocol, in cases where a person has been recognised as not having the capacity to consent, be he or she a minor or an adult, a genetic test may be carried out on that person only for his or her direct benefit.

90. The second paragraph provides some clarifications concerning minors. In such case, a genetic test shall be deferred until the person has attained the capacity to consent. However there is provision for making an exception to this if carrying out the test without delay is in the minor's best interests. In particular this covers cases where delaying the test might harm the subject's health by depriving him or her from a health benefit. For example, when the information which the genetic test would provide would allow appropriate therapeutic measures to be taken for a disease or disorder from which the subject is suffering.

91. The exception also applies to some situations where the genetic test would provide predictive information allowing timely preventive measures to be taken. In particular this applies to tests for diseases which might develop before the subject has attained legal capacity.

92. In addition to the consequences on the subject's health of deferring the test, regard is also had to the subject's well-being in deciding whether to perform a test without delay. The word "well-being" refers both to physical and mental well-being. For example, a predictive test performed without delay and producing a negative result might spare the subject highly invasive regular examinations as in the case of adenomatous polyposis.

93. For adults not able to consent, where the clinical situation of the person makes it possible, it is good practice to defer genetic tests until recovery of capacity to consent.

### **Article 11 – Information prior to authorisation, genetic counselling and support**

94. Article 10 of this Protocol provides that subject to Article 13, a genetic test may only be carried out on a person who does not have the capacity to consent, for his or her direct benefit. Compliance with this requirement is therefore an essential precondition for envisaging a genetic test on a person who does not have the capacity to consent.

95. In accordance with Article 6 paragraph 4 of the Convention, Article 11 provides in its paragraph 1 that the person, authority or body whose authorisation is required for a genetic test on a person not able to consent, must be given appropriate information beforehand. This information is the same, in both content and form, as that which must be given in the case of persons able to give consent, under Article 8.

96. The article also requires that appropriate prior information is provided, to the extent of his or her capacity to understand, to the person not able to consent on whom a test is envisaged. Capacity to understand must be construed in a relatively broad sense as taking in both discernment and reasoning. In the case of a minor such capacity will depend, in particular, on the subject's age and degree of maturity.

97. The person, authority or body whose authorisation is required and, where appropriate, the person concerned by the envisaged test, may wish certain information concerning the test to be further clarified. In the case of a person being able to consent, these clarifications can be obtained by the person him or herself, within the framework of his or her dialogue with the doctor, in view of an informed decision. Such is not the case for the person, the authority or body whose authorization is required for a test on a person not having the capacity to consent. It is important that a suitably trained person with the necessary knowledge of clinical genetics be available to answer these possible questions. The aim is to facilitate and ensure understanding of the information provided.

98. The provisions of Article 8 paragraph 2 concerning genetic counselling also apply to persons not able to consent when their capacity to understand permits this. For predictive tests referred to in Article 12 of the Convention, the person concerned shall therefore, where possible, have access to appropriate genetic counselling.

99. The last sub-paragraph provides that, where appropriate, appropriate follow-up be available for the person whose authorisation is required, in order to allow him or her to deal with the implications of the test and its results. Such requirement is explained in particular by the predictive nature of information obtained by the envisaged test and the implications for the person on whom the test is being considered and, where appropriate, for the members of his or her family.

#### **Article 12 - Authorisation**

100. In accordance with paragraphs 2 and 3 of Article 6 of the Convention, this article lays down the principle that genetic tests may not be carried out on persons not able to consent, whether minors (paragraph 1) or adults (paragraph 2), without authorisation from their representative, or authority, person or body provided for by law. However, to preserve their autonomy to the maximum extent possible:

- minors' opinions must be taken into consideration as an increasingly determining factor, in proportion to their age and degree of maturity;
- whenever their capacity to understand permits this, adults must be involved in the authorisation process.

101. The second sub-paragraph in paragraph 2 applies to adults who have foreseen the possibility of becoming not able to give consent, and have indicated their wishes concerning a genetic test in advance. In accordance with Article 9 of the Convention, those wishes shall be taken into account.

102. Because of the predictive nature of the results of the tests referred to in paragraph 2 of Article 8 and their potential implications for the person concerned and/or the members of his or her family, paragraph 3 provides for the authorisation to be documented. As a general rule, the authorisation will be given in writing and will bear the signature of its author. This does not rule out the possibility of using, in certain cases, other forms of registration of authorisation as long as it permits to identify without ambiguity the person on whom the test is envisaged and the person, authority or body giving the authorisation.

103. Paragraph 4 provides that the person, authority or body who gave the authorisation may withdraw it at any time provided that this is in the best interests of the person not able to consent. In this regard, as for the authorisation process, the latter's opinion shall be taken into

consideration, in the case of a minor as an increasingly determining factor in proportion to his or her age and degree of maturity and, in the case of an adult, depending on his or her capacity to understand.

## **CHAPTER VI – Tests for the benefit of family members**

### **Article 13 - Tests on person not able to consent**

104. This article lays down the conditions to be met so that exceptionally and by derogation from the provisions of Article 6 of the Convention on Human Rights and Biomedicine and Article 10 of this Protocol, a test may be carried out on a person not able to consent without direct benefit for him or her.

105. The purpose of the test must be to enable family members, with whom the person concerned has a biological link, to obtain a preventive, diagnostic or therapeutic benefit that has been independently evaluated as important for their health, or to allow them to make an informed choice with respect to procreation.

106. The situation in question can be illustrated by three examples. The first corresponds to the case of a person not able to consent and suffering from cancer, which is considered to have a genetic basis. The performance of a genetic test identifying the involved genetic mutation may not modify the way in which the cancer of this person will be treated. But, the test could provide information which could be used for the analysis of the genetic characteristics of the other members of the family in order to determine if they are likely to develop the same cancer. If the same mutation is found in some of the family members, they could be subject to more frequent controls in order to allow cancer detection at an early stage of the emergence of the disease.

107. Another example is that of a child who was diagnosed, on the basis of clinical signs and symptoms and biochemical tests, with cystic fibrosis. This disease can be related to many different genetic mutations. For possible future procreation choices, it can be important to identify the existing mutation in the affected child. This will make it possible to look for the mutation in the child's parents in order to determine if it is them who transmitted it or if it is a mutation newly appeared in the child having developed the disease. In the latter case, there would be no particular reason to fear for the health of a future child of the couple concerned.

108. A last example is the case of diseases, especially rare ones, for which the genetic mutation involved has not been identified. In such case, the transmission of the mutation can be traced by studying genetic linkage. In order to determine a genetic risk in a family in which a genetic disease with an unidentified genetic mutation has manifested itself, it is possible that genetic tests on affected but also unaffected children would be necessary, so as to obtain an acceptable degree of diagnostic certainty – for example, for other members of the family, whether of child-bearing age or not.

109. This evaluation of the benefit for the family members must not be made by the medical doctor of the family members for the benefit of whom the test is envisaged. However, it is not required to set up a special structure, and the choice of entrusting for example another medical doctor or a body (e.g. committee, tribunal, etc.) with the task of carrying out the evaluation is left to the State.

110. The second condition is that the benefit envisaged can only be obtained by performing the test. The term "benefit envisaged" refers here both to the nature of the benefit and to the time it takes to materialise. The benefit envisaged must be necessary now for the health of a (the) family member(s) and, in particular, cannot wait, when the test is to be carried out on a minor, for the minor to reach legal capacity. This provision would also be considered to cover the case where the information sought for the benefit of a (the) family member(s) could be obtained without carrying out a test on the person not able to consent but only at the cost of highly complex and laborious analyses.

111. Paragraph c lays down another condition: the risks and burdens associated with the intervention shall be minimal for the person undergoing the test. This means that the intervention would only result at the most in a very slightly detrimental and temporary impact on the health of the person on whom the test is carried out. Furthermore, the expected discomfort, which might be associated with the intervention, shall be at most temporary and very slight for this person. Thus taking blood samples from peripheral vessels will be considered as acceptable.

112. Paragraph d. requires that the expected benefits of the test be independently evaluated as substantially outweighing risks for private life that may be associated with collection, processing or communication of the data it produces. These data are the main concern.

113. In accordance with Article 16 of this Protocol, personal data resulting from a genetic test are considered confidential. Their collection, processing and communication must meet the requirements of the legislation governing the protection of individuals with regard to the processing of personal data. In the cases covered by this article, the genetic test is carried out on one person but in the context of an analysis involving one or more members of his or her family, who are informed of the test and will benefit from its results. Even if such cases are very rare, provision must be made for these particular situations where the exchanges of information within the family concerned can be made easier. Paragraph d takes into account the risks, arising in particular from the very special situations involved, to the private life of the person on whom the test is being considered. It requires that the expected benefits have been independently evaluated as substantially outweighing those risks. The clarification concerning “independent evaluation” in paragraph a are also relevant here.

114. Authorisation by the representative of the person, on whom the test is to be carried out, or by an authority, person or body provided for by law, is also required. In the specific case of this type of test, as the representative of the person not able to consent is often a member of his or her family, he or she may also be a beneficiary of the test envisaged. The law should make proper provision for such situations.

115. Finally, with a view to respecting, as far as possible, the autonomy of person on whom the test is to be carried out, he or she must be associated with the authorisation procedure, as far as his or her capacities to understand and degree of maturity permit.

116. In any case, if he or she objects, the test must not be carried out. This last provision reiterates the requirement already stated in the specific field of biomedical research and transplantation of organs and tissues, in Articles 17 and 20 of the Convention and the additional corresponding Protocols. The concept of objection implies from the person concern a certain capacity to understand and that he or she expresses a will. A difference must be made between a simple gesture of fear or dissatisfaction by a young child, gesture which is not to be termed legally as an objection, and the will expressed by an older child capable of discernment. However, even with a young child with no capacity of discernment, attention shall be paid to the requirement that the medical intervention does not entail more than minimum burden.

#### **Article 14 – Tests on biological materials when it is not possible to contact the person concerned**

117. This article concerns genetic tests envisaged on biological material of a person for the benefit of his or her family members. Under Article 9 of this Protocol, the consent of the person concerned must be obtained for all genetic tests. This also applies to tests carried out on biological material originally removed from that person for another purpose. Reasonable efforts, in terms of both means and time, must therefore be made to re-contact the person on the biological material of whom the test is considered.

118. However, in spite of such reasonable efforts, in some rare cases, it may prove impossible to get in contact with the person. Whereas in some cases, failure to carry out the envisaged genetic test may have serious consequences for the health of those family members whom it was intended to benefit. This applies, for example, to families in which there have been several cases of ovarian cancer, and the genetic mutation involved has not been identified. The genetic test envisaged might help to carry out a family study with a view to identify that mutation, making it unnecessary to remove the ovaries of female family members in whom it would not be identified. In such cases, it may be considered that the benefit for the family members concerned is particularly important and substantially outweigh any risks to the person whose biological material would be used – in particular for his or her private life – if the test were carried out without his or her consent.

119. In the case when it is not possible to get in touch with the person concerned, this article provides that the law can, in accordance with the principle of proportionality, allow the test to be carried out under condition that the expected benefit cannot be otherwise obtained and that the test cannot be deferred.

120. The objective of the test must be to allow family members to obtain an important benefit for their health in terms of prevention, diagnosis or treatment, or enable them to take informed decisions in relation to procreation choices.

121. In accordance with the principle of necessity, the expected benefit cannot be obtained without the test being carried out and the test cannot be deferred. Furthermore, in accordance with the principle of proportionality, the expected benefit must be significantly greater than the risks to the individual's private life that may arise from the collection, use or communication of the test results.

122. Finally, the article requires that provisions be made, in agreement with Article 22 of the Convention, for the cases where the person concerned has expressly objected to such a test. Article 22 of the Convention enacts the general rule according to which the parts of the body removed with a given purpose can only be stored and used for other purposes if done in conformity with appropriate information and consent procedures. In this respect, the Explanatory Report to the Convention specifies that information and consent arrangements may vary according to circumstances. In respect of these principles it is the responsibility of each state to put in place the necessary provisions to address the situations where an objection would have been expressly made by the person concerned.

#### **Article 15 - Tests on deceased persons**

123. The possibility of carrying out a genetic test on biological material removed from the body of a deceased person, or removed, when he or she was alive, from a person now deceased, can prove to be important from a medical point of view, for the members of his or her family. However, a legal framework for such a possibility must be defined.

124. The article requires that the consent or authorisation required by law be obtained before carrying out any genetic test on biological samples removed from the body of a person after his or her death for the benefit of family members of this person. The same requirement applies to test performed on biological material removed, from a person, when he or she was alive and is now deceased.

125. It is left to national law to determine the rules governing consent (e.g. express or presumed) or authorisation applicable to genetic tests hence implemented.

126. It is also left to national law to specify the conditions for the evaluation of “the benefit of other family members”. For the evaluation of such a concept, the principles of necessity and proportionality must be taken into account.



127. In the field of genetics, because of the biological links with the deceased person, descendants or other family members may have specific interests. Even though, according to law, the decision is not for them to take, they should be consulted, taking into account in this context the risk of conflict of interest regarding the benefit envisaged.

## **CHAPTER VII – Private life and right to information**

### **Article 16 - Respect for private life and right to information**

128. In accordance with Article 10 of the Convention on Human Rights and Biomedicine, Article 16 recognises the right to respect for private life, when it comes in particular to personal data derived from a genetic test. It thus reaffirms the principle laid down in Article 8 of the Convention for the Protection of Human Rights and Fundamental Freedoms, and re-stated in the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data.

129. The genetic tests covered by this Protocol aim at providing information concerning health. It should be pointed out that, under Article 6 of the Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data, personal data concerning health constitute a special category of data regarded as particularly sensitive, and require thus appropriate guarantee.

130. However, a genetic test performed for health purposes may incidentally provide personal information not relating to health which could have important consequences for the person concerned or members of his or her family. For example it might bring to light the default of an expected biological link or an unexpected presence of a biological link.

131. The conditions of form and substance for making such unexpected information available to the person concerned are a matter for national law. In this connection, regard should be had, in particular, to any wishes expressed by the person concerned about having access to such information, and regard should likewise be had to any damage which such information might bring to this person or other members of his or her family.

132. Paragraph 2 gives everyone the right to know all information collected on his or her health by a genetic test. The individual in question must therefore have in principle access to all the data that concerns him or her, contained in particular in his or her medical file.

133. On the other hand, with regard to the conclusions of medical nature drawn from the test, the article lays down a further requirement: the person concerned must have access to these in a form that is comprehensible. Where necessary, this could require the conclusions to be reworded in more simple terms so that they can be understood by the individual in question, in the light of his or her level of knowledge, education and psychological state. In certain cases, like those of the tests covered by the exceptions envisaged in Article 7 paragraph 2, the dialogue with the medical doctor will not be possible; other arrangements will have to be envisaged in order to fulfil the requirements of paragraph 2.

134. The right to know goes hand in hand with the right not to know as recognised in paragraph 3. The person may have his or her own reasons for not wishing to know about certain aspects of his or her health. This may apply, for example, to a test aiming at identifying the presence of a gene responsible for a particularly serious disease, for which there is currently no treatment. The person concerned may agree to the test – the results of which could be useful for procreation choices or for the health of members of his or her family – without wanting to know whether he/she carries that gene. This wish must be observed.

135. Both the right to know and the right not to know may, in specific circumstances, be subject to certain restrictions in the interests of the person concerned. Information on the health of a person who has expressed a wish not to know is sometimes particularly important for him or her. For example, knowing that he or she has a predisposition to a disease might

be the only way to enable him or her to take measures to prevent that disease or delay its development. In such cases, the doctor's duty to provide care, stated in Article 4 of the Convention on Human Rights and Biomedicine, might conflict with the patient's right not to know. It is up to national law to indicate whether, having regard to the circumstances of the particular case, the doctor may make an exception to the right not to know.

#### **Article 17 – Biological samples**

136. The biological samples on which genetic tests referred to in Article 2 of this Protocol are carried out, shall only be used and stored in such conditions as to ensure their security. Appropriate measures shall thus be taken in particular, against accidental or unauthorised deterioration, destruction as well as unauthorised access or use.

137. Biological samples may, moreover, be a source of information on the persons from whom they have been removed. Samples must therefore be used and stored under such conditions as to ensure the confidential nature of the information which might be obtained from them.

#### **Article 18 – Information relevant to family members**

138. The results of certain genetic tests may be relevant for the health of other family members. The person concerned must have been informed of this before the test is carried out, in accordance with the provisions of Article 8 of this Protocol.

139. When analysis of the test results confirms the relevance for the health of his or her family members, the person on whom the test has been carried out must be informed and made aware of the importance of access to this information for his or her family members. The person should be made aware of this at an appropriate time, depending, amongst other things, on his or her clinical situation and any decisions on his or her health that could be taken in the light of the test results.

140. For the communication of this information to the family members, appropriate provisions should be made, bearing in mind the rules on confidentiality and the protection of the private life of the various persons concerned (person on whom the test is performed and members of his or her family). The choice of procedure(s) is left to the States. If the person tested is unable or unwilling to contact his or her family members directly he or she may be given appropriate material or letters to pass on to the family member(s). Consideration could be given to setting up a mediating body responsible for contacting family members of the person concerned if the latter has asked for them to be informed without him or herself being identifiable as the source of the information. Another example, would be the possibility to provide for a decision by a competent body, following comparative assessment of the respective interests of the persons concerned, on whether or not the information in question must be communicated to the members of the family.

141. The persons informed of the importance for their health of the results of a test performed on a member of their family should be invited to consult a medical doctor and, where a genetic test referred to in Article 8 paragraph 2 is envisaged, appropriate genetic counselling shall also be available for them.

### **CHAPTER VIII – Genetic screening programmes for health purposes**

#### **Article 19 – Genetic screening programmes for health purposes**

142. Article 19 states the conditions to be fulfilled before implementing a genetic screening programme. "Screening programme" refers to the general offer of a genetic test to an entire population or to a section of population, with a view to enable early identification or exclusion of:

- a genetic disease,
- a genetic predisposition to a disease, or
- another genetic characteristic relevant for the health of the members of this population or section of population or for their descendants (e.g. a gene involved in resistance to a disease).

143. Genetic tests proposed as part of a screening programme are motivated by a public health concern. Their relevance for each member of the population or section of population concerned is not based on individual or family data. It differs in this respect from the relevance of a test carried out in the context of medical supervision, which is based on individual indications. The purpose of proposing a genetic test as part of a screening programme for health purposes is to allow the members of the population or section of population concerned to make appropriate personal choices concerning their health or in relation to procreation, on the basis of the results of the proposed test.

144. The approval by a competent body is required before the implementation of a screening programme for health purposes. The term "competent body" means a body recognised by the State to perform a certain task.

145. The approval by this body may only be given subject to the fulfilment of specific conditions, which must be independently evaluated.

146. The specific conditions listed in sub-paragraphs a. to d. of this article supplement those defined in the other chapters of this Protocol which apply at individual level, in particular the general provisions (Chapter II) as well as those concerning the quality of genetic services and the clinical utility of genetic tests (Chapter III), information, genetic counselling and consent (Chapter IV) and private life and right to information (Chapter VII).

147. The programme must be recognised for its health relevance for all the members of the population or section of population concerned. The disease targeted by the screening must represent a significant health problem for the population concerned because of its severity or the number of people affected. The programme will enable the members of the population concerned to take appropriate decisions, in particular concerning the prevention or treatment of the disease.

148. The scientific validity of the screening programme and its effectiveness must have been established. This scientific validity must be determined in respect of the purpose of the screening and on the basis of the sensitivity, specificity and reliability of the proposed test which must fulfil the requirements of paragraph a. of Article 5 of this Protocol. A reduction in mortality or morbidity is generally considered an essential criterion of the effectiveness of a screening programme.

149. To that end, appropriate preventive or treatment measures must be available to the persons concerned. The notion of prevention is to be understood here in a broad sense, including in particular close health monitoring, for example through regular examinations (e.g. for a predisposition to breast cancer). This may include measures (as in the case of cystic fibrosis or Duchenne's muscular dystrophy, for example), that limit or delay the development of the disease symptoms and that significantly improve the well-being or living conditions of the person concerned. Finally, a choice concerning procreation – a field which is relevant to health – may, in certain cases, in particular when there is a risk of transmission of a particularly severe disease very difficult to treat, be considered as an appropriate measure.

150. Carrying out a pilot study on a small section of the target population before implementing the programme will make it possible to evaluate all the negative and positive implications of the proposed programme at every level: technical, organisational, scientific, clinical and even psychological and social.

151. Appropriate measures shall be provided to ensure equitable access to the screening programme to the members of the population or section of the population concerned. This provision reiterates the principle established in Article 3 of the Convention applied here to screening. Equity in this context primarily means the absence of discrimination. Subsequently, not being synonymous with absolute equality, the equitable access indicates a satisfactory degree in access to screening.

152. Lastly, the programme shall provide for measures to allow the population or section of the population concerned to benefit from adequate information concerning the screening programme. The information shall enable the persons concerned to know the existence of the screening programme and its purpose, as well as the voluntary nature of participation in it. It shall also specify the way to access to the programme and where to obtain more detailed information, in particular on the screening process, benefits and possible negative effects (for example, anxiety regarding the test results) and possible implications for family members, so as to enable them to decide whether or not to accept the test proposed.

## **CHAPTER IX – Public information**

### **Article 20 – Public information**

153. The purpose of the article is to prompt the States to take appropriate measures to facilitate access for the public to objective general information about genetic tests. Genetics is increasingly going to become an integral part of health care. However it is still a relatively new and complex field. Outside a medical context the possibilities of access to objective general information about genetic tests are still often very limited. To enable better understanding of this area of genetics, the progress it opens up for health and its limitations, it is important for the general public to have access to such information in a comprehensible form about the different types of test available (diagnostic tests, tests predictive of monogenic diseases and those permitting to detect a predisposition to a disease, healthy carrier tests). They must also relate to the applications of these tests in the medical field and in particular to the extent and, if necessary, the limit of the significance of the information resulting from such tests. Such information is also contributing to the prevention of stigmatisation based on genetic characteristics.

154. The choice of appropriate measures is left to the individual State; and will depend in particular on the quality of the information already accessible for the public. Information campaigns or creation of Internet sites are examples of how to meet the object of informing the general public. Education and training syllabuses could also be added to for that purpose. Promoting and supporting such initiatives are examples of measures which States can take to satisfy the requirements of this provision.

155. Such general information could, when appropriate, be supplemented by other more specific information on particular tests, in particular tests available outside individualised medical supervision. The companies marketing such tests often advertise them extensively, and may not always provide the information which the persons concerned need for making an informed choice. In this context, availability of objective information about such tests is particularly important and would help to protect the public against incomplete, wrong or in some cases deliberately misleading information.

## **CHAPTER X – Relation between this Protocol and other provisions and re-examination of the Protocol**

### **Article 21 - Relation between this Protocol and the Convention**

156. As a legal instrument, the Protocol supplements the Convention. Once in force, the Protocol is subsumed into the Convention for those Parties having ratified the Protocol. The provisions of the Convention are therefore to be applied to the Protocol.

157. Thus, Article 36 of the Convention, which sets out the conditions under which a State may make a reservation in respect of any particular provision of the Convention, will also apply to the Protocol. Using this provision States may, under the conditions set out in Article 36 of the Convention, make a reservation in respect of any particular provision of this Protocol.

#### **Article 22 – Wider protection**

158. In pursuance of this article, the Parties may apply rules of a more protective nature than those contained in the Protocol. In other words, the text lays down common standards with which States must comply, while allowing them to provide greater protection of the human being and of human rights with regard to genetic testing for health purposes.

#### **Article 23 - Re-examination of the Protocol**

159. This article provides that the Protocol shall be re-examined no later than five years from its entry into force and thereafter at such intervals as the designated Committee may determine. Article 32 of the Convention identifies this Committee as the Steering Committee on Bioethics (CDBI), or any other Committee so designated by the Committee of Ministers.

### **CHAPTER XI – Final clauses**

#### **Article 24 – Signature and ratification**

160. Under the provisions of Article 31 of the Convention, only States that have signed or ratified the Convention may sign this Protocol. Ratification of the Protocol is subject to prior or simultaneous ratification of the Convention. A State which has signed or ratified the Convention is not obliged to sign the Protocol or, if applicable, to ratify it.