68/CNECV/2012

NATIONAL COUNCIL OF ETHICS FOR THE LIFE SCIENCES

OPINION ON THE DRAFT DECREE-LAW THAT REGULATES LAW NO. 12/2005 OF 26 JANUARY, WITH REGARD TO GENETIC INFORMATION, GENETIC DATABASES AND GENETIC TESTING

(November 2012)

DRAFT PROPOSAL ON THE REGULATION OF GENETIC TESTING
(Opinion on the Draft Decree-Law that regulates Law no. 12/2005 of 26 January, with regard to genetic information, genetic databases and genetic testing)

I – INTRODUCTION

An opinion was requested of the National Council of Ethics for the Life Sciences (CNECV) on the draft 'decree-law [that] sets out the principles inherent to the performance and availability of genetic tests, and also providing the rules for protection of genetic information, in terms of access, security, confidentiality and secrecy of data. Thus regulated are no. 6 of article 6, no. 2 of article 7, no. 1 of article 15 and no. 7 of article 17 of Law no. 12/2005 of 26 January'.

II – GENERAL FRAMEWORK

The CNECV issued Opinions on previous occasions, namely Opinions no. 43/CNECV20/04 – Opinion on Draft Law no. 28/IX – personal genetic information and health information; and no. 56/CNECV/2008 – Opinion on direct sale to the public of genetic testing.

The CNECV considers essential, from an ethical point of view, that the principles of primacy of person and search for human well-being be explicit; the principles of respect for autonomy (that arises from the freely given informed consent and proper counselling); of equity in access to health care; respect for confidentiality and privacy (implying the protection of information, professional confidentiality, privacy, and safety of the means, resources and equipment); as well as non-discrimination and non-stigmatization. In turn, the safety and quality of the services and procedures are also ethical benchmarks to take into account.

III – ANALYSIS OF THE LEGISLATIVE PROJECT

According to the Preamble of the Draft, it recognises the need for regulation of no. 6 of article 6 (genetic information), of no. 2 of article 7 (genetic databases), of no. 1 of article 15 (laboratories carrying or offering genetic testing), and no. 7 of article 17 (protection obligation) of Law no. 12/2005 of January 26. Regulation on these matters and the initiative to carry it out is considered relevant. Which may, today, allow to rethink aspects of the 2005 standards.

The document in question has some strengths, particularly the protection of the consent and the subject's data and the safety-related aspects of genetic information; it also has innovative points, such as the national genetic network or the separation of genetic information in the computer systems and access thereto. But it needs to be refined and clarified in some articles, as it seems unrealistic, particularly within the prescribed period of adaptation.

Moreover, Law no. 12/2005 of 26 January, that this decree regulates in four articles, is probably of the most restrictive in terms of information and prescription of medical acts – for example, by establishing that genetic testing for detection of heterozygous state for recessive disorders, presymptomatic diagnosis of monogenic diseases and genetic susceptibility testing in healthy people *can only be carried out (...) at the request of a physician specialized in genetics* (no. 2 of Article 9), imposes an unprecedented restriction. This Council believes that this point should be reviewed.

We will now appraise the proposal of this draft.

Chapter I – General provisions

The issue of definitions is not unimportant. Some of the proposed terms or expressions lack refinement – an example of this is paragraph a) of article 3, whereby, instead of 'sick individual or at risk' it should be 'sick person or at risk and, where appropriate, the family'.

The principles[1] presented in the general provisions seem connected to 'topics' and the wording could be of greater conceptual rigour[2]. For example, in paragraph a) of no. 1 of article 4, it should be 'primacy of the person' instead of 'primacy of the patient'; in paragraph b), 'equity in the access to health care' instead of 'equal access of the citizens to health'; in paragraph f), instead of 'evidence', the 'use of the best scientific practices, based on evidence'.

Chapter II – Creation of genetic databases

[1] These are relative to: 'reception, processing and transmission of genetic information, as well as the availability, performance, interpretation and marketing of genetic tests' (no. 1 of article 4); 'creation, processing and access to genetic information and genetic databases'(*ibid.*, no. 2 and 3) and the primacy of the 'rights and interests of the holder of genetic information' (*ibid.*, no. 4). In the following chapters, there are more principles, operationalized, for example, in the requirements for creating databases (consent, strictly used for the purpose consented to, revocation at any time), in the methods for collecting genetic information (principle of physical and moral integrity and risk minimisation), in data quality ('principles of legality, transparency, protection of personal identity, of the best scientific practices and good faith; to be necessary, adequate, relevant and not excessive in relation to the purposes for which it is provided; to be exact ' – article 12, no. 1).

[2] In no. 2 of article 4, one must consider 'principles of respect for human dignity and autonomy of the person from whom the information results and the consent as well as confidentiality and privacy' (instead of considering as principles 'the preservation of private and family life, of informed consent and confidentiality of personal data').

The *Requirements for setting up databases* (article 5) associate the following cumulative conditions: the need for setting it up, the consent, the favourable opinion of the Medical Genetics Committee (CGM) and the notification to the National Data Protection Commission (CNPD). It would be necessary to clarify the competences proposed to the CGM and notify the CNPD; indeed, it is considered more appropriate to request favourable opinion to the CNPD and not just notification.

With regard to the consent (article 8) that binds the processing of personal genetic information to the intended purposes (in article 6), the form written by the person him/herself or his/her legal representative (if the data owner lacks capacity), makes revocation at any time clear, without disadvantages to the person him/herself. We consider we have to highlight the figure of consent, 'depending on their age and their level of maturity and ability to understand', to be obtained in the case of minors and those that are incapable. Even so, their involvement 'insofar as possible' in the process of decision making is vague and there would be gains in using the terms of the Convention on Human Rights and Biomedicine, to clarify that their opposition should be respected or, at least, that they had not expressed their opposition.

The terms of the consent that appear in article 9 (Right to information) give rise to the need for clarification of the terms in its no. 1. Since the issue of information and consent is crucial, we warn that, in practice, the person responsible for the information or its owner may not be who more adequately conveys the information to its owner.

The combined analysis of articles 8 (Consent) and 10 (Right of opposition from data owner) gives rise to a redundancy between no. 4 and 5 of article 8 and no. 1 and 2 of article 10, the first being defined more incipiently than in article 10. Indeed, there could be the interpretative gains in putting no. 6 and 7, referring to revocation, as the new no. 3 and 4 of article 10.

Chapter III – Database Maintenance and Management

The person responsible for genetic information has to ensure the maintenance and supervision of the genetic database and section I decrees that: 'Whenever the database is intended for or used for the provision of health care, the person responsible for the genetic information should be a specialist doctor in medical genetics.'

According to the definitions set forth in paragraph l) of no. 1 of article 3, the 'Person responsible for the genetic information' means the individual appointed by the data processing head as the one responsible for the genetic information. Hence, it is not understandable that the person responsible for the genetic information has to be a specialist in medical genetics, since clinical practice in this

area is not required for such functions. Moreover, other qualified professionals, duty bound to confidentiality, could perform these functions to the benefit of the processes and the people. Using the definition given for 'laboratory geneticists' in paragraph e) of no. 1 of the same article, the professionals able to pursue the laboratory practice of medical genetics shall be 'doctors, biologists and pharmacists with specialization in genetics conferred by the respective professional associations, senior health technicians of the genetics branch and professionals of related biomedical areas with training in human genetics and specific laboratory training and recognised officially to carry out genetic testing related to health.'

Section II – Communication, Interconnection and Cross-Border Flow of personal genetic information

No. 2 of article 16 of the Draft says that 'it is forbidden to copy to all kinds of external information support any personal genetic information contained in the genetic database', which can create difficulties when, exceptionally, to request a diagnostic test or for a particular treatment, it is necessary to have access to certain genetic information without which the diagnostic test and/or the therapy may not be the most appropriate. Thus, seeking the balance with clinical usefulness, we suggest the following: '2 – It is forbidden to copy to all kinds of external information support any personal genetic information contained in the genetic database, *unless such information is crucial for the purposes of medical diagnosis, the provision of care or treatment to the owner.*'

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Chapter IV – Protection of personal genetic information

Access to genetic information is processed differently according to their medical or non-medical nature and their purpose – to provide health care or biomedical research. In no. 2 and 3 of this article 21, it makes little sense to limit so severely the access 'to doctors responsible for health care to be provided or provided to the owner of the information', considering that there is intervention by health professionals subject to professional regulations and involved in the process of genetic counselling. If we consider that such genetic studies are performed in laboratories, the aforesaid professionals also have access to personal genetic information. It is suggested that the wording of the article considers the effective presence of the professionals involved in providing care to that person or family. In no. 4 of the same article, the text relating to obtaining information 'freely and without constraint at reasonable intervals and without delay or excessive cost' is formulated vaguely, by the use of the terms *reasonable* and without *excessive* cost.

In no. 3 of article 22, the CNECV considers the statement that 'the computer systems should ensure the logical separation between genetic information, personal data and clinical data, and include different levels of authorized access' to be relevant.

Chapter V – Performance and provision of genetic testing

Section I – Medical genetics laboratories

As for the wording of article 25, the technical management, besides the mentioned profile, should include the profiles of laboratory geneticists (paragraph e) of article 3); it is not clear if the personnel requirements apply to existing staff (since it requires that the minimum qualification is a degree and three years experience) or to new contracts; and the need to mention 'deontological standards' is questionable, as this would require that each of the professions involved had defined the content of its professional deontology.

Article 27 gives details of the 'Administrative procedure for the recognition of a benchmark medical genetics laboratory' that are unclear: the reason for the Directorate-General of Health (DGS) to check the 'respective geographic scope', and the fact that the criteria '*may be* implemented by order of the member of Government responsible for health': for the benefit of rigour, the criteria not only *can* but *should* be operationalized (no. 6).

Section II – Genetic testing

The responsibility 'for conducting the necessary studies in populations, (...) by presenting evidence of analytical and clinical validity of the tests offered and the analysis of their clinical usefulness,' lies with the 'medical genetics laboratories': it seems to us that who effectively determines the mentioned analytical and clinical validity is not explained.

As for the Subcontracting of tests (article 31), certain aspects do not seem to have been taken into account, such as response time or the cost of the test, or if there is a study of a family member as index-case analyzed in a laboratory duly accredited by Standard ISO 589.

We consider, in relation to direct sales and promotion, that the principles pointed out in specific CNECV Opinion – Opinion 56/CNECV/2008 on Direct Sale to the Public of Genetic Testing – have been complied with.

Chapter VI – Genetic counselling

The Draft provides for access to medical genetics consultation, the creation of the national referral network and genetic counselling. It is important to emphasise the ethical concern with people, their well-being, and training to deal with any difficult situations or of high uncertainty, of their own or of their families.

Chapter VII – Medical Genetics Committee

Considering the purpose of this Committee – 'to provide technical advice and assist in the formulation of policies and implementation of programmes and other initiatives in the field of human genetics and health' – and its generic multidisciplinary composition – 'non-medical and medical specialists of recognised merit in clinical, laboratory and research field, appointed by order of the Member of Government responsible for health' –, the designation of this entity should mirror its composition, so it is suggested that it should be called Human Genetics Committee.

In the Competences provided for in article 41, these stand out: the evaluative [a), g), j)]; of proposal [*b), c), d), e), f)];* and of consultation [*h, i), l); l)*]. Accordingly, the Committee receives requests for opinions, in the field of human genetics and health, from the Director-General of Health or the member of Government responsible for the area of health; however, the possibility of requesting a CNECV Opinion should also be expressed.

According to no. 4 of the same article, the Committee 'may, if necessary, seek the assistance or advice of ethics committees for health pursuant to Decree-Law no. 97/95 of 10 May'. However, such a measure is not provided for in the regulation of the Ethics Committees for Health[3] (CES), regulations that should be updated.

Chapter VIII – Sanctions Regime

In article 42 of the Draft, in the administrative offences are not included (i.e. specially regulated) the violations of articles 6, 9, 10, no. 2 of article 13, article 15, 19, no. 1 to 3 of article 20, 23, 25, no. 1 of article 30 – so that, according to no. 3, 'the legal regime for administrative offences approved by Decree-Law No. 433/82 of 27 October is applicable'. It is suggested that this situation be reviewed considering at least the non-compliance of articles 9, 12 and 23.

Despite being provided for in no. 2 of article 43 (additional sanctions) that the 'failure to comply with the provisions of article 14 determines the application of the additional sanction of prohibition of sale of genetic testing', the time is not formulated (or the minimum and maximum range) of the prohibition. On the other hand, it cannot be understood why the sale of genetic tests in breach of article 14 (Destruction or disruption of the database) is forbidden, but is not, though it should be, in

[3] In Decree-Law No. 97/95 of 10 May, which regulates the Ethics Committees for Health, in accordance with article 6, it shall be the responsibility of the CES: 'b) Issue, on its own initiative or upon request, opinions on ethical issues in the field of activities of the institution or the respective health service'; and, in no. 1 of article 7, 'May request the CES to issue opinions: a) The management bodies of the respective health service or institution; b) Any health care professional of the respective health service or institution; c) Patients or their representatives, through the Board of Directors of the institution or health service.'

the case of non-compliance with article 36 (Conservation, protection and destruction of biological material used in genetic testing).

With regard to article 44 (Joint and several liability), the CNECV questions the grounds for considering the genetic testing laboratory referred to in article 14 (Destruction or disruption of the database) or in no. 1 of article 20 (regarding the responsibility of the person responsible for the treatment) jointly and severally responsible. In the event of offence, the responsibilities should be investigated and the offender punished.

Chapter IX – Transitional and final provisions

The 180-day period provided for in article 47 of the Draft for adaptation of genetic laboratories (public and private) that already exist may be insufficient, given the need for a quality management process for a non-certified institution. In addition, there is news regarding the computer system, according to no. 3 of article 22, as well as the creation of the national network. We think that this period should be extended to actually be feasible.

IV – CONCLUSION AND RECOMMENDATIONS

Accordingly, taking into account the principles laid down in the framework and bearing in mind that we recognize an ethical value in the promotion of coherent legislation, adjusted to the needs of health and protection of the rights of citizens, the National Council of Ethics for the Life Sciences is of the opinion that this Draft Decree-Law:

1. Whilst not raising fundamental ethical objections, can however be improved, both in terms of accuracy and in terms of procedures and procedural aspects related to the protection of the person and their follow-up and support; in this sense, there are some reservations regarding the clarity of the document as was pointed out in the course of this Opinion.

2. It is found wanting on what happens to existing databases, set up without term of consent for the treatment of genetic information – to accompany all requisitions/requests for tests and bearing the names of the owners, tests ordered and respective results. It is reasonable to conclude that all existing laboratories have ‘genetic databases’ as a requirement for its proper functioning, and that, also for the conservation of genetic information, they will depend on the consent, even if later, of the owners of the information. However, this

consent may be unenforceable but, on the other hand, the destruction of samples seems undesirable as it would impair further research.

3. The CNECV recommends a reflection on the advantages for citizens and for the interprofessional activities, of a review of Law No. 12/2005 of 26 January, in what it most restrictively presents, in particular the wording of no. 2 of article 9, with regard to the monopoly for requesting genetic testing by medical geneticists.

Lisbon, 19 November 2012

The President,
Miguel Oliveira da Silva

This Opinion was approved in the plenary meeting of 19 November 2012. Besides the Chairman, the following Counsellors were present:

Lucília Nunes (Rapporteur); Agostinho Almeida Santos; Carolino Monteiro; Isabel Santos; José Germano de Sousa; José Lebre de Freitas; Lígia Amâncio; Michel Renaud; Pedro Nunes.