Definitions of genetic testing in Italian legal documents

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In their interesting paper “Definitions of genetic testing in European legal documents,” Varga et al. (2012) provide an extensive assessment of how European and other legislation and policy instruments define genetic testing. They found that the definitions used across documents lacked a general consensus, showed that a definition of genetic testing was more often provided in non-binding than in binding instruments, and emphasized that a differentiation needs to be made between clinical genetic testing, genetics laboratory-based genetic testing, and genetic information.

Overall, the authors analyzed 59 legal documents; one of which is a policy instrument by the Italian National Bioethics Committee (NBC) (1999). However, four further Italian legal documents (two policy papers and two binding instruments) define genetic testing. We thus provide a brief description of these documents and of the definitions for genetic testing they set forth and compare them with the findings of Varga et al., with the aim of contributing to a more complete and accurate view of how European legislation and policy instruments define genetic testing.

The first policy instrument we refer to is the 1998 paper by the Italian National Committee for Biosecurity, Biotechnology and Life Sciences (NCBBS) entitled “Guidelines for Genetic Testing” (Italian National Committee for Biosecurity 1998). The aim of this paper was to provide a reference framework to healthcare and laboratory professionals involved in genetic

testing, with the overall purpose of ensuring that genetic testing is safe and effective, is used appropriately, is performed in accordance with high quality standards, and is offered to users whose decisional autonomy is fully respected and who are given appropriate social and psychological support.

The second policy instrument is the 2011 position paper “Genetic Susceptibility Testing and Personalized Medicine” (Italian National Bioethics Committee and Italian National Committee for Biosecurity and Life Sciences Joint Working Group 2010) by the NBC/NCBBS joint working group. The aim of this paper was to provide the general public with a critical assessment of the current and potential value of genome-wide testing in terms of clinical validity and clinical utility.

Of the two binding instruments, the first is an agreement reached in 2004 by the Italian Ministry of Health and regional health authorities, the “Guidelines for Medical Genetics Activities” (Permanent Conference of the State and the Regions and the Autonomous Provinces of Trento and Bolzano 2004), which regulates medical genetics services. This binding instrument mainly focuses on the organization of clinical medical genetic services and laboratories, on the requirements that such facilities must meet, and on the training of medical genetic professionals.

The second binding document is a regulation that is periodically issued by the Italian Data Protection Authority, the “General Authorization for the Processing of Genetic Data” (Italian Data Protection Authority 2011). The main purpose of the authorization is to establish the purposes for and mechanisms whereby genetic data can be processed. It is a soft-law instrument that would typically lack binding force but benefits from a hybrid status, as the Italian Data Protection Code confers such force to provisions dealing with genetic information (Republic of Italy 2003). In its current version (Italian Data Protection Authority 2011), the authorization sets forth an extensive list of definitions for genetic testing and related concepts (Battistuzzi et al. 2012). We have analyzed the definitions of genetic testing laid out in these documents

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Table 1 Items covered by the definitions of genetic testing contained in Italian policy and legislation documents

Document	Legal field		Type of testing (context)											
	Labor/insurance law	Privacy, confidentiality, and biobanks law	Health care law	Forensic/criminal/civil identification/family law	Complex	Diagnostic testing	Presymptomatic testing	Disease predisposition testing	Drug response	Carrier testing	Prenatal testing	PGD	Population screening	Identity testing
Guidelines for Genetic Testing	+	+	+	+		+	+	+		+	+		+	+
Guidelines for Medical Genetic Activities			+			+	+	+	+	+			+	+
Genetic Susceptibility Testing and Personalized Medicine	+		+			+	+	+	+	+			+	+
General Authorization for the Processing of Genetic Information	+	+	+	+		+	+	+	+	+			+	+
Document														
	Object (material/method)													
	Chromosomes			DNA			Specific gene products			Routine blood tests			Family history	
Guidelines for Genetic Testing	++			++			++							
Guidelines for Medical Genetic Activities	++			++			++							
Genetic Susceptibility Testing and Personalized Medicine	++			++			++							
General Authorization for the Processing of Genetic Data	++			++			++							

+ “legal field” covered by each document; presence of a given term or phrase (context)

++ presence of a given term or phrase

according to the criteria applied by Vargas et al.; the results are shown in Table 1.

We observed that none of the Italian documents, including those that are specifically concerned with data protection, personal rights, and privacy, mention family history or routine laboratory tests as sources of genetic information. Interestingly, the analysis of Varga et al. shows that roughly a third of the European documents (21/59) include family history, and only nine mention routine laboratory tests, as “materials” or “methods” from which genetic information can be obtained. It has been suggested that definitions associated with genetic information often center on genetic testing because it may be easier to specify and control due to its more recent and less well-established medical and social role, as opposed to family history or routine tests (Parthasarathy 2004). However, because family pedigrees can be important indicators of genetic risk and genetic information can be derived from nongenetic laboratory tests, the absence of these contexts of application from the definitions of genetic testing contained in the Italian instruments we identified, as well as from most of the European documents reviewed by Varga et al., seems to reflect an incomplete understanding of what the term genetic testing may encompass.

Another result worth pointing out has to do with prenatal genetic testing and preimplantation genetic diagnosis (PGD). Prenatal testing is included as one of the possible types of testing in the definition of genetic testing provided by just one of the Italian documents. The others discuss prenatal testing, but place it under the wider umbrella of diagnostic and presymptomatic testing. As for PGD, it is completely absent from all of the Italian instruments. According to the review of Varga et al., just over half (34/59) of the European documents’ definitions of genetic testing include either PGD or prenatal testing. In the case of PGD, this may be explained by the fact that its legal status has seen numerous, often controversial, developments, both in Italy (Levi Setti et al. 2011; Republic of Italy 2004) and in other European countries such as Germany (Tuffs 2011). Prenatal testing, however, is one of the most widely employed clinical applications of molecular genetics. That it is not mentioned as one of the settings for genetic testing in so large a subset of the legal instruments discussed here indicates that widespread difficulties exist in the use of appropriate, well-defined terminology.

Overall, despite the shortcomings described above, we found that the four Italian documents clearly distinguish between clinical and nonclinical genetic testing, take into account the setting for clinical applications, and discriminate between diagnostic testing and the range of predictive tests

conducted in healthy individuals. Most importantly, their definitions of genetic testing show a good degree of consensus, providing a framework that appears to be both coherent and enforceable (Sequeiros 2010). These results contrast with the conclusions of Varga et al. that the definitions set forth in the European instruments they surveyed suffered from extreme variability, and sometimes displayed a lack of clarity and precision, and point to how eliminating language barriers and providing international accessibility of national hard- and soft-law instruments would allow for more accurate analyses of legal data coming from different sources.

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