The Open Public Health Journal

ISSN: 1874-9445 — Volume 12, 2019

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RESEARCH ARTICLE

Genetic Research: The Role of Citizens, Public Health and International Stakeholders

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Abstract

Background:

Genetic research has become an indispensable instrument for medical research, and the subjects involved have both divergent and convergent interests.

Objective:

The possibility of having more detailed genetic information undoubtedly offers benefits for the health of the subject, but could also pose risks and make the subject vulnerable to discrimination.

Methods:

The scientific community has viewed very favorably the public health utility of family history, in which data from a family whose members suffer from chronic pathologies is collected and filed, in order to develop a sort of “stratification of family risk.”

Even though in the last decade the scientific and juridical literature has contributed greatly to the topic of biobanks, the perplexities that continue to surround this theme give the idea that current ethical protocols on research are inadequate.

Results:

Researchers, citizens, International stakeholders, mass media, Public Health and Governments play a key role in genetic research. It is obvious that the methods used for genetic research do not present intrinsic risks; they are much less dangerous than other activities of diagnosis and research. Before authorizing a research project, it is important to reflect on the responsibility and transparency of the studies to be conducted, and on the impact they may have on the interests of public health.

Conclusion:

We believe that the highest priority need is to develop a common language on the theme, as is the case in the sphere of clinical experimentation where rules of good clinical practice, albeit at times conflicting, have led to uniform convergences in the scientific world on the points to be actuated.

Keywords: Biobanks, Genetics research, Role of citizens, International stakeholders, Public health, Chronic pathologies.

Article Information

Identifiers and Pagination:

Year: 2019
Volume: 12
First Page: 106
Last Page: 113
Publisher Id: TOPHJ-12-106

Article History:

Received Date: 30/09/2018
Revision Received Date: 12/02/2019
Acceptance Date: 10/03/2019
Electronic publication date: 28/03/2019
Collection year: 2019

Article Metrics:

CrossRef Citations: 0

Total Statistics:
Full-Text HTML Views: 133
Abstract HTML Views: 107

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1. INTRODUCTION

DNA is “the deepest and most essential patrimony of the human person” [1], because it is shared with other people from the same geographical area. It may predict future events or the possibility that may occur. It is easy to obtain, and can be of interest to third parties [2] such as family members, insurance companies and employers.

Therefore, there is great interest in this information, espe cially since genetic predisposition has been clearly demons trated for various diseases such as cardiovascular pathologies [3, 4] diabetes [5], late onset Alzheimer [6], schizophrenia [7] bipolar disorder [8], autism [9], cancer [10-12] and rare diseases [13].

The collections of tissues derived from human bodies and used for the extraction of genetic material have become known under various names such as biobanks, biolibraries, tissue repositories, genetic databases, or DNA banks.

The OECD defined human biobanks and genetic research databases as ‘structured resources that can be used for the purpose of genetic research, which includes human biological materials and/or information generated from the analysis of the same; and extensive associated information [14]

The purpose of genetic biobanks [15] is to support research to identify the mutations that cause genetic diseases, since they permit researchers to collect material from families affected by the same pathology, with an enormous saving of energy and funds.

There has been an acceleration in the study of "complex diseases" [16] whose etiopathogenesis involves both genetic and environmental factors, as the existence of biobanks has enabled greater comprehension of pathogenetic mechanisms, development of new diagnostic instruments and the design of treatment strategies [17]. The enormous number of samples available has made it possible to identify a distinction between "susceptibility" and "genetic causality."

Another particularly fertile field for genetic research is that of chronic pathologies. The *Evaluation of Genomic Applications in Practice and Prevention* (EGAPP) [18], created in 2004 by the *CDC National Office of Public Health Genomics* in the United States, has studied the genetic aspects of many tumoral pathologies by referring to samples in biobanks, with good applicative outcomes.

At the same time, when biobanks facilitate the study of various factors of genetic risk related to an illness, they potentially influence the societal conceptions of responsibility, group identity and future options [19].

In fact, new quandaries emerge when genetic research identifies groups of subjects who carry genes that place them at higher risk for a disease. These "individuals at risk" fall into a disease category that is ambiguous because of the slippage between the risk factor and the disease itself [20]: they are not ill, but may become so. How should they be treated? As sick people? As healthy people at risk? Should they be counselled not to have children? Should they be subjected to higher health insurance premiums? Epistemic change influences cultural change, and in this case can lead to the creation of a cultural current that views genetic research with suspicion.

In the scientific literature, genetic exceptionalism [21, 22] is the line of thought according to which all the information derived from DNA, because of the intrinsic characteristics of the genetic patrimony, must be considered a separate category from the common information that can be deduced from in-depth family anamnesis, and should be afforded particular protection.

The need for genetic privacy [23] is fueled by the fear of discrimination, social stigmatization, family problems, loss of control of one's identity, as well as the psychological implications, because genetic information can be "potentially embarrassing and uniquely personal" [24].

This issue entails not only political but even more importantly ethical and social choices, because the fundamental rights of the person are involved, as well as individual and collective interests.

2. METHODS

In this work, we will discuss the role that researchers, physicians, ethics committees, citizens, public health officials and international stakeholders can play in the promotion and control of genetic research that draws upon biobanks. We will examine the current situation in Italy and compare it with that of other Western countries.

3. RESULTS AND DISCUSSION

3.1. Physicians/Investigators (And Research Ethics Committee)

stores and distributes quality DNA, cell and tissue samples for scientists conducting research on rare diseases genetic material and rare diseases" [26].

However, recent years have seen a growing awareness that transnational collaboration is essential researchers have access to a greater quantity of biological samples. It is no coincidence that in the recent years there has been an increase in the number of transnational associations and entities involved with genetics, which permit the circulation of ideas in various affiliated nations. An example is the P3G Consortium [27], an international not-for-profit organization that catalogues experiences in the field of population genetics, in order to build common research strategies to facilitate harmonization and open the door to future collaborations. Another valid example of openness in this sense is the pan-European Biobanking and Biomolecular Resources Research Infrastructure - European Research Infrastructure Consortium (BBMRI-ERIC [28]), whose funding includes a grant from the European Unions' Horizon 2020 research and innovation program.

In this context, it is noteworthy that in multicentric studies, data are often collected and then transmitted to third parties.

That operate in different nations, which in turn have different sets of legislation on the subject, but this by no means exempts those who receive this data from responsibility for the correct handling of the information. It is extremely important that the personal data remain the exclusive prerogative of the research, and not be exploited for purposes other than the research itself.

The scientific community has viewed very favorably the public health utility of family history [29], in which data from a family whose members suffer from chronic pathologies is collected and filed, in order to develop a sort of "stratification of family risk."

These few examples help us understand how essential it is to expand the borders of genetic research to improve public health.

It is obvious that the methods used for genetic research do not present intrinsic risks; they are much less dangerous than other activities of diagnosis and research. However, there is an "information risk" for the patient [30], who may suffer psychological harm from receiving the genetic test results themselves or from the way the results are conveyed, or for the patient's relatives, since the results may have repercussions for them as well.

In order to deal with these ethical and legal implications, North American organisations, followed by European Union ones, have published guidelines or ethical codes that serve genetic professionals on an international scale. Another issue to be faced is the problem of incidental findings discovered in the course of the research. The committees of the Clinical Sequencing Exploratory Research (CSER) Consortium and the Electronic Medical Records and Genomics (eMERGE) Network [31] asserted that researchers must limit themselves to informing participants about the research results; they have no moral obligation to undertake clinical tests on the basis of these results, according to the principle that clinical research is distinct from medical care in both its aims and its guiding moral principles. However, physicians are required to provide complete information expressed in terms that the subject can understand. In addition, once the information has been obtained from the genetic tests, physicians will tend to give more or less importance to certain data on the basis of their scientific and cultural formation.

Obviously, the problem is complicated in the case of anonymous samples: on the one hand, donors cannot be informed if a genotype at risk is revealed, but on the other hand, they do not run the risk that their genetic data may be misused.

Anonymization of data in the constitution of biobanks is of particular importance because it entails the irreversible loss of the connection between personal data and genetic information. The connection constitutes the added value of biobanks. When biobanks are established, the decision whether to anonymize data or not is of crucial importance. When the connection between personal data and genetic

the identity of the donor of a sample is an added value, as donors can be informed of findings that may prove crucial to their health and well-being.

The first to grasp the importance of this aspect were the Scandinavian nations. For example, a 1999 document of the "Swedish Medical Research Council [32]" defined biobanks as collections of human tissue samples, the origin of which must always be traceable. Italy lacks a national law on the subject, but the 2016 Authorisation for the use of genetic data prepared by the Guarantor for the Treatment of Personal Data states that the treatment of genetic data is allowed only for purposes of prevention, diagnosis or treatment of the subject, or of scientific research, or for purposes of proof in civil or penal cases, according to the dictates of law.

Thus Italy allows scientific research on genetic material, as long as it respects the rules set forth in the Authorisation for the use of genetic data. Instead, there is no specific legislation on the establishment and use of biobanks.

Of central importance in this legislative vacuum are the ethics committees [33], which evaluate the ethics of studies on genetic data, and also express opinions on research programs that involve the study of data or samples when for particular reasons it is not possible to inform the donors [34-36].

Serious vigilance on the part of the Research Ethics Committee and self-control on the part of researchers are the basis for credible genetic research that respects human dignity.

3.2. The Role Of Citizens

The possibility of having more detailed genetic information undoubtedly offers potential benefits for the health of the subject, but could also pose risks and make the subject vulnerable to discrimination in cases in which the genotype is used to draw conclusions about the phenotype.

Early knowledge about one's own genetic characteristics and the probability of contracting a pathology provides the basis for taking preventive measures. In the case of a multifactorial disease, that is, one in which there is an association between environment and genotypic characteristics, steps can be taken to prevent the pathology by adopting changes in lifestyle, diet, work and the environment itself.

But knowledge of genetic information can change an individual's self-perception and deeply influence the character of his or her social organization, in cases in which the genetic patrimony could be an obstacle to a certain type of work. There could also be significant implications for family planning. *For example*, if a couple is aware that one of them has a predisposition to a genetic disease, they may choose not to have children [37]. Or when Non-Invasive Prenatal Testing (NIPT) are performed, information about the fetus could induce a disproportionate and unjust recourse to abortion in nations that permit it, becoming a form of biological eugenics [38, 39] or of "positive eugenics [40]", the goal of which is to avoid unfavorable medical conditions [41], rather than impose a genetic structure on future generations [42].

According to the Denver Post [43], 80 to 90 percent of women who receive a positive result from an amniocentesis test for Down Syndrome choose to terminate the pregnancy. Their decision is not made on the basis of a moral evaluation [44], but is grounded in concern about how having a Down Syndrome child will affect their life as a couple.

Clearly, knowledge about the fetus' state of disability can give the couple greater awareness of their situation as parents, whether they

It is fundamental that citizens be well informed, so that they will not make extreme choices that lack grounding in valid science, or worse, refuse to consider genetic testing to understand their health problems.

The scientific community has been reflecting for many years on the need to inform citizens adequately about the issue of genetic research. In this regard, numerous international studies have explored people's awareness about the risks of genetic engineering, their rights to receive or give information (including the responsibility to family and society), and their involvement in public debate on genetics. One example is the "European COB [45]" - Challenger of Biomedicine, Meetings and Minds.

The committees of the Clinical Sequencing Exploratory Research (CSER) Consortium and the Electronic Medical Records and Genomics (eMERGE) Network [46] assert that participants in research, if they have given informed consent, must be informed of the results of a study and participate in an appropriate clinical follow up.

It is evident that directly involved citizens, such as those suffering from neurodegenerative pathologies or cancer [47], are more likely to donate their tissues and support the establishment of biobanks designed to study their pathology, while the average citizen for whom this issue has no relevance, or members of ethnic or religious minorities, tend to be reluctant to participate in genetic studies.

While it is commonly accepted that a patient who participates in a genetic study must be thoroughly informed, it is not so obvious that average citizens involved in population studies' must be educated about the new technologies and their use, or the risks and benefits to the community they may pose

This issue came to international attention in 1996, when Icelandic citizens gave explicit consent for an initial collection of DNA and implicit consent for a second one for the deCODE Genetics [48] study, which also gathered sensitive health-related data archived in the nation's Health Sector Database. There was international debate on the importance of transparent provision of information so that citizens are fully aware of the ramifications of their involvement.

Since 2005, numerous conferences on the subject have been organized with the participation of the community of scientists and bioethics specialists, and representatives of organizations of patients. The conclusions drawn at these gatherings have stressed the need for developments in legislation and monitoring systems. They have also emphasized the importance of avoiding pressure from economic interests and providing equal access to treatment. They have stated that freedom of choice is paramount, and have discussed the decision-making powers of ethics committees.

They have pointed out that the lack of public participation in the debate regarding new genomic technologies [49-51] highlights a deficit in Western democracies, while, conversely, active involvement promotes social justice, confirming Kant's view that all people and their points of view on the issue are important [52].

If the national healthcare system were able to identify families or entire populations that have a predisposition to certain genetic diseases, it could establish prevention and early treatment programs for these categories. If, instead, citizens do not wish to participate in genetic screening programs, the healthcare authorities will not have the data they need for identifying these predispositions, and consequently the subjects at risk will not benefit from actions the authorities might have been able to undertake to identify and treat these categories of at-risk citizens. By now it is clear that patients need to take on a key role in genetic research [53], not only because doing so directly provides material for studies, but also because they can influence their governments' choices about the research programs to be undertaken to improve the general health of the nation.

could happen. Therefore, while today it is impossible to answer questions about distributive justice, it is nonetheless important that civil society be aware of these issues and observe future developments in biobank-based research with an eye to this important aspect.

Citizens who make available their own body or health data when they participate in epidemiological studies or research on a disease can benefit from the opportunity to know the biological characteristics of their own state of health. Just as important, their choice demonstrates the importance of social solidarity, because their entire community benefits from the knowledge acquired from the study. In fact, citizenship in a community is a source of rights, but also of responsibilities toward that community.

The duty of social solidarity was emphasized by the Universal Declaration on the Human Genome and Human Rights, adopted unanimously by UNESCO in 1997 [52, 54], which is the first document of universal importance in the field of bioethics. It was written to provide ethical and legal principles for the promotion of freedom of research, human dignity, solidarity and international cooperation.

Subjects who voluntarily participate in genetic research or who are invited to participate, because of clinical reasons or for purposes of statistics, must be informed about the consequences of having a genetic test done or not having it done.

Social solidarity may strongly influence the decision of citizens to participate in genetic research in the interests of benefit-sharing. As stated in the Declaration on the Human Genome [55] (1997), “Benefits from advances in biology, genetics and medicine, concerning the human genome, shall be made available to all, with due regard for the dignity and human rights of each individual. Freedom of research, which is necessary for the progress of knowledge, is part of freedom of thought. The applications of research, including applications in biology, genetics and medicine, concerning the human genome, shall seek to offer relief from suffering and improve the health of individuals and humankind as a whole (art. 12)”.

The HUGO Ethics Committee [56] (2000) approved a declaration on the sharing of benefits from genetic studies, which should not only have positive effects for the health of subjects involved in the research, but also should provide broader and more immediate gain for these communities in terms of investments in welfare by private firms that benefit financially from the samples donated.

The HUGO [57] Statement on human genomics databases (2002) also indicates that these biobanks should be “global public goods” and that there should be “fair and equitable” distribution of the benefits of research. It also called for recognition of the rights of researchers, institutions and business entities to a “fair return” for their intellectual and financial contribution (recommendation 6).

On the basis of social solidarity and the duty of subsidiarity of citizens who participate in genetic research, some authors [58-60] have proposed using biological samples for future research even without specific informed consent, or when there is a generalized consent that allows any kind of future research without specifying the details. In this way, citizens would not benefit personally from the research but could contribute to the common good in terms of public health.

Others go much farther, perhaps in doing so undermining social solidarity. Some authors have proposed dynamic consent, obtained through the use of new computer technologies to reach patients [61]. This method of acquisition of consent, through a digital communication interface, facilitates two-way communication to stimulate a more engaged, informed and scientifically literate participant population where individuals can tailor and manage their own consent preferences.

Regardless of the method used to obtain informed consent, it is evident that studies on genetic material have significant ethical-legal and social reverberations, and thus great caution is required in governing access to the data, as well as in controlling how it may be made public.

genotypes, for whom bulk drugs are ineffective: these minorities of individuals with particular genotypes would end up not receiving drugs specifically for them.

3.3. Mass Media

In this context, according to the Council of Europe [64], the mass media play an important role in the spread of information about genetics, and are key to promoting the citizen participation in the discussion on the human genome.

In the case of the UK Biobank and that of the Islandic deCODE Genetics project, the mass media encouraged the legitimacy of the existing research infrastructures.

3.4. Public Health And Governments

Since the early 1990s, the establishment of biobanks has been considered an attractive economic activity [65] because the results of scientific research based on biobank samples could serve in drug development, could inform disease prevention policies, and could be useful to insurance corporations in designing and adapting policies. The problem is that this economic aspect can undermine the social solidarity that motivates the donation of these tissues from which genetic data can be obtained. Economic interests could come to outweigh the needs of medical science, and thus the benefits of the results obtained would not be shared with the very populations who were the object of the studies.

The potential alliance of public healthcare policies and genetic research depends on choices currently being made.

The task at hand is certainly difficult because entire chapters of the education and perception of scientific knowledge must be re-written, with the unavoidable emergence of new responsibilities (who should manage a national biobank, and how should it be run?)

A new approach is needed for the classic themes of ethics such as informed consent and data security, as well as autonomy and privacy (either in an existential sense or as a practical problem of confidentiality [66])

In our opinion, a government-run national biobank would best protect the interests of the people who donated tissues.

The literature on the theme provides no uniform interpretations of the role of genetics in public health: many countries tend toward total interference of research in the life of citizens (*e.g.*, Denmark [67, 68], Belgium [69], Iceland, and Australia [70]) while others have taken a more prudent approach, that is, they have not created national infrastructures, but, as in the case of Italy, have many small collections at public or private institutes.

The difference between the two approaches is substantial. When a national government acquires and uses genetic data from its citizens, storing this information in a national databank, an individual's genetic information can serve health of the entire national community. Instead, when genetic data is stored and used by a number of many small public or private institutes, an individual's genetic information can serve his or her family and a few involved individuals, such as spouses.

Clearly, in both choices, it is important to identify new responsibilities and opportunities, not only those gained but also those lost [71].

may arise. Instead, we think it important to promote the culture of the establishment of a biobank to obtain useful results for families and all of society.

The greater the number of samples collected in a national biobank, the more feasible it is for genomic analysis to move from scientific, clinical, governmental and commercial settings to that of personalized genomic medicine for the nation's citizens. However, what will happen if "stratified medicine" [71] becomes "stratified markets"? When the potential sales to certain subgroups or entire nations are too insignificant to merit investment by pharmaceutical firms, these people could end up lacking the "personalized genomic medicine" that could cure their health problems. Would this social injustice be addressed by their governments, which, after all, promoted a national biobank of its citizens' biological samples?

Before authorizing a research project, it is important to reflect on the responsibility and transparency of the studies to be conducted, and on the impact, they may have on the interests of public health.

3.5. International Stakeholders

The theme of the human genome has been addressed in terms of respect for human dignity and the fundamental rights of the person by numerous international documents, such as the Recommendation of the Committee of Europe regarding genetic data and tests [72], the 1977 Convention on Human Rights and Biomedicine, the 1997 UNESCO Universal Declaration on the Genome and Human Rights, the EU's 2000 Charter of Fundamental Rights, the November 1996 Code of Conduct of the International Labour Organization on the protection of the personal data of workers, the Helsinki Declaration of the World Medical Association (June 1964 and successive modifications), and the European Commission Working Document on Genetic Data adopted March 17, 2004 by the Working Party for the Protection of Individuals with regard to the processing of personal data.

The Treaty on the Functioning of the European Union (TFUE), in requiring institutions and organisms of the EU, as well as the member states, to respect the free circulation of personal data in the exercise of activities that have to do with the application of the law of the Union (art. 16) also established that "independent authorities" (instituted by law CE/2001/45 e and reasserted by the new Regulation (UE) 2018/1725) should supervise this.

Even though in the last decade the scientific and juridical literature has contributed greatly to the topic of biobanks, the perplexities that continue to surround this theme give the idea that current ethical protocols on research are inadequate [73].

Now, as never before, international organizations have a crucial role in delineating the route for correct integration of genetics in public health.

Stakeholders can influence the government and its changing public healthcare policy, but coordination between them at various levels is fundamental.

It would be interesting to produce an internationally recognized ethical-legal code of good practice concerning the use of biological samples, one that would enable all researchers wherever they work to gain access to biobanks in the same way, and that would establish the same technical regulations for the organization of the biobanks. Another idea could be a "free zone for research," where the same technical regulations would attain, and where the maximum protection of human dignity would be ensured. Furthermore, it would be interesting to establish a European observatory on genetic studies, or even a world-level one, similar to the observatory on clinical experimentation of

CONCLUSION

Genetic research has become an indispensable instrument for medical research, and the subjects involved have both divergent and convergent interests.

On the one hand, the public wants research to advance so that gains can be made for the health of everyone and benefits can be shared, but at the same time, people are frightened by the risk of possible discriminatory uses of the information acquired. On the other hand, researchers and research institutes call for greater incentives for their work, appealing to the principle of solidarity, and at the same time demand the rights to exploit the intellectual property associated with their discoveries.

There are also the international stakeholders whose excessive protection of information risks are slowing or even paralyzing scientific progress.

Finally, there are the governments of the individual nations who, digging in and waiting for greater clarification on the theme, have failed to legislate appropriately or have abdicated their role to technical organisms (*e.g.*, the Guarantor in Italy), without resolving the problem, except in a sectorial way.

In this Babel of overlapping and at times conflicting interests, one risks losing sight of the objective: personalized medicine that can truly help patients.

Genetic data must be used not to exploit, but to serve the person. Freedom and responsibility must be the twin guiding lights for establishing parameters for the use of biological samples. An evaluation of how this technology impacts the various aspects of the future of society is urgently needed.

ETHICS APPROVAL AND CONSENT TO PARTICIPATE

Not applicable.

HUMAN AND ANIMAL RIGHTS

No animals/ humans were used for the studies that are the basis of this research.

CONSENT FOR PUBLICATION

Not applicable.

CONFLICT OF INTEREST

The authors declare no conflict of interest, financial or otherwise.

ACKNOWLEDGEMENTS

All individuals listed as authors have contributed substantially to the design, performance, analysis, or reporting of the work and are required to indicate their specific contribution. Nunzia Cannovo has contributed to the drafting, Piergiorgio Fedeli has written the article, Claudia Casella and Caludio Buccelli have substantially contributed important intellectual content to the study, and Vincenzo Graziano has contributed in revising the manuscript.

REFERENCES

[1] Piazza A. Le biobanche: tra genetica e diritto. Iustitia 2006; 1: 41.

[2] Cannovo N, Paternoster M, Buccelli C. Predictive genetic tests for employment purposes: why not? Med Law 2010; 29(3): 419-32. [PMID: 22145561]

[3] Wang L, Fan C, Topol SE, Topol EJ, Wang Q. Mutation of MEF2A in an inherited disorder with features of coronary artery disease. Science 2003; 302(5650): 1578-81.[http://dx.doi.org/10.1126/science.1088477] [PMID: 14645853]

[4] Savoia C, Volpe M, Grassi G, Borghi C, Agabiti Rosei E, Touyz RM. Personalized medicine-a modern approach for the diagnosis and management of hypertension. Clin Sci (Lond) 2017; 131(22): 2671-85.[http://dx.doi.org/10.1042/CS20160407] [PMID: 29109301]

[5] Florez JC. Mining the genome for therapeutic targets. Diabetes 2017; 66(7): 1770-8.[http://dx.doi.org/10.2337/dbi16-0069] [PMID: 28603140]

[6] Myers A, Holmans P, Marshall H, *et al.* Susceptibility locus for Alzheimer's disease on chromosome 10. Science 2000; 290(5500): 2304-5.[http://dx.doi.org/10.1126/science.290.5500.2304] [PMID: 11125144]

[7] Sundby A, Boolsen MW, Burgdorf KS, *et al.* Stakeholders in psychiatry and their attitudes toward receiving pertinent and incident findings in genomic research. Am J Med Genet A 2017; 173(10): 2649-58.[http://dx.doi.org/10.1002/ajmg.a.38380] [PMID: 28817238]

[8] Kennedy JLA, Farrer LA, Andreasen NC, Mayeux R, St George-Hyslop P. The genetics of adult-onset neuropsychiatric disease: complexities and conundra? Science 2003; 302(5646): 822-6.[http://dx.doi.org/10.1126/science.1092132] [PMID: 14593167]

[9] Monaco AP, Bailey AJ. Autism. The search for susceptibility genes. Lancet 2001; 358(Suppl.): S3.[http://dx.doi.org/10.1016/S0140-6736(01)07016-7] [PMID: 1178 4552]

[10] Peto J. Cancer epidemiology in the last century and the next decade. Nature 2001; 411(6835): 390-5. [http://dx.doi.org/10.1038/35077256] [PMID: 11357148]

[11] Ragin C, Park JY. Biospecimens, biobanking and global cancer research collaborations. ecancer 2014; 8: 454.

[12] Falco G, Rocco N, Bordoni D. Contralateral risk reducing mastectomy in Non-BRCA-Mutated patients. Open Medicine (Poland) 2016; 11(1): 238-41.[http://dx.doi.org/10.1515/med-2016-0089]

[13] Krier JB, Kalia SS, Green RC. Genomic sequencing in clinical practice: applications, challenges, and opportunities. Dialogues Clin Neurosci 2016; 18(3): 299-312.[PMID: 27757064]

[14] Organisation for Economic Co-operation and Development. Guidelines for human biobanks and genetic research databases Paris, OECD 2008. Available from: http://www.oecd.org/sti/ emerging-tech/guidelines-for-human-biobanks-and-genetic-research-databases.htm [Accessed on January, 2019]

[15] SIGU TELETHON Linee guida per le Biobanche Genetiche 2003. Available from: www.sigu.net [Accessed on July 21, 2018]

[17] Presidenza del Consiglio dei Ministri, Comitato Nazionale di Bioetica, Comitato Nazionale per la Biosicurezza, le Biotecnologie e le Scienze della Vita 2009. Available from: http://presidenza.governo.it/ biotecnologie/ [Accessed on July 21, 2018]

[18] Evaluation of Genomic Applications in Practice and Prevention (EGAPP). Available from: www.cdc.gov/egappreviews/default.html [Accessed on July 21, 2018]

[19] Lindgaard Hoeyer K. Size matters: The ethical, legal, and social issues surrounding large-scale genetic biobank initiatives. Nor Epidemiol 2012; 21(2): 211-20.

[20] Greene JA. Prescribing by Numbers: Drugs and the Definition of Disease 2007.

[21] Otlowski M. Exploring the concept of genetic discrimination. J Bioeth Inq 2005; 2(3): 165-76.[http://dx.doi.org/10.1007/BF02448597] [PMID: 16578943]

[22] Murray TH. Genetic exceptionalism and “Future Diaries”: Is genetic information different from other medical information?Genetic secrets: Protecting privacy and confidentiality in the genetic era 1997.

[23] Everett M. Can you keep a (genetic) secret? The genetic privacy movement. J Genet Couns 2004; 13(4): 273-91. [http://dx.doi.org/10.1023/B:JOGC.0000035522.58496.c8] [PMID: 19736694]

[24] Annas GJ. Privacy rules for DNA databanks. Protecting coded ‘future diaries’. JAMA 1993; 270(19): 2346-50. [http://dx.doi.org/10.1001/jama.1993.03510190102034] [PMID: 823 0598]

[25] Elger B, Biller-Andorno N, Mauron A, Capron AM. Ethical Issues in Governing Biobanks: Global Perspectives 2008.

[26] http://www.eurobiobank.org/ [Accessed on December 28, 2018]

[27] Cannovo N, Folliero V, Russo A, Meccariello E. The role of international stakeholders in genetic research 2013.Proceeding of the 9th World Conference on Bioethics, Medical Ethics & Health Law 2013 November 19-21; Naples, Italy. 2013.

[28] European Research Infrastructure Consortium. Ethical, Legal and Societal issues http://www.bbmri-eric.eu [Accessed on December 28, 2018]

[29] Yoon PW, Scheuner MT, Peterson-Oehlke KL, Gwinn M, Faucett A, Khoury MJ. Can family history be used as a tool for public health and preventive medicine? Genet Med 2002; 4(4): 304-10.[http://dx.doi.org/10.1097/00125817-200207000-00009] [PMID: 1217 2397]

[30] Di Lorenzo P, Casella C, Capasso E, *et al.* The central importance of information in cosmetic surgery and treatments. Open Medicine (Poland) 2018; 13(1): 153-7.[http://dx.doi.org/10.1515/med-2018-0024. eCollection 2018]

[31] Jarvik GP, Amendola LM, Berg JS, *et al.* Return of genomic results to research participants: the floor, the ceiling, and the choices in between. Am J Hum Genet 2014; 94(6): 818-26.[http://dx.doi.org/10.1016/j.ajhg.2014.04.009] [PMID: 24814192]

[32] Abott A. Sweden sets ethical standards for use of genetics biobanks. Nature 1999; 400(July): 13.[PMID: 10403239]

[33] Ricci G, Cannovo N. La. 2010.2010.Proceedings of the FIRB Conference 2010, June 3-4; Ferrara, Italy. 2010.

[34] Art. 110. Legislative Decree "Codice in materia di protezione dei dati personali" 30th June 2003, n. 196, publish on Official Gazzette n. 174 of 29th July 2003

[35] Art. 11.5, Legislative Decree “Codice in materia di protezione dei dati personali “- Disposition of the Garante n. 2 of the June 16th 2004 (Official Gazzette August 14th 2004, n. 190)

[36] art. 8 of the “Autorizzazione al trattamento dei dati genetici” February 22nd 2007.

[37] Paternoster M, Saccone G, Maruotti GM, *et al.* Ethical challenges in pregnant women with brain injury. Journal of Maternal-Fetal and Neonatal Medicine 2018; 31(17): 2340-1.[http://dx.doi.org/10.1080/14767058.2017.1339271]

[39] Piras M, Delbon P, Bin P. Voluntary termination of pregnancy (medical or surgical abortion): Forensic medicine issues. Open Medicine (Poland) 2016; 11(1): 321-6.[http://dx.doi.org/10.1515/med-2016-0062]

[40] Ricardo MLB, Roberto JBB. Is genetic counseling a form of eugenics? Rev Cienc Salud 2018; 16(1): 10-26.

[41] Ravitsky V. The shifting landscape of prenatal testing: Between reproductive autonomy and public health. Hastings Cent Rep 2017; 47(Suppl. 3): S34-40.[http://dx.doi.org/10.1002/hast.793] [PMID: 29171889]

[42] Casella C, Capasso E, Terracciano L, *et al.* Ethical and legal issues in gestational surrogacy. Open Medicine (Poland) 2018; 13(1): 119-21. [http://dx.doi.org/10.1515/med-2018-0019]

[43] The rise of a new eugenics. https://www.denverpost.com/ 2013/07/03/the-rise-of-a-new-eugenics/

[44] Gillott J. Screening for disability: A eugenic pursuit? J Med Ethics 2001; 27(Suppl. 2): ii21-3. [http://dx.doi.org/10.1136/jme.27.suppl_2.ii21] [PMID: 11574654]

[45] Institut fur Ethik und Geschichter der Medizin. Challenges of Biomedicine –Socio-Cultural Contexts, European Governance, and Bioethics Available from: http://www.egmed.uni-goettingen.de /index.php?id=183 [Accessed on July 21, 2018]

[46] Jarvik GP, Amendola LM, Berg JS, *et al.* Return of genomic results to research participants: the floor, the ceiling, and the choices in between. Am J Hum Genet 2014; 94(6): 818-26.[http://dx.doi.org/10.1016/j.ajhg.2014.04.009] [PMID: 24814192]

[47] Malone T, Catalano PJ, O'Dwyer PJ, Giantonio B. High rate of consent to bank biologic samples for future research: The Eastern Cooperative Oncology Group experience. J Natl Cancer Inst 2002; 94(10): 769-71.[http://dx.doi.org/10.1093/jnci/94.10.769] [PMID: 12011228]

[48] Merz JF, McGee GE, Sankar P. "Iceland Inc."?: On the ethics of commercial population genomics. Soc Sci Med 2004; 58(6): 1201-9. [http://dx.doi.org/10.1016/S0277-9536(03)00256-9] [PMID: 1472 3914]

[49] Burgess MM, Tansey J. Democratic deficit and the politics of "informed and inclusive" consultation.Hindsight to Foresight in Emerging Technologies 2008; 275-88.

[50] Dodds S, Ankeny RA. Regulation of hESC Research in Australia: Promises and Pitfalls for Deliberative Democratic Approaches. Bioethical Inquiry 2006; 3: 95-107.[http://dx.doi.org/10.1007/s11673-006-9007-4]

[51] Gottweis H. Emerging forms of governance in genomics and post-genomics:structures, trends, perspectives.Genetic Governance: Health, Risk and Ethics in the Biotech Era 2005.

[52] Foltz F. Five arguments for increasing public participation in making science policy. Bull Sci Technol Soc 1999; 19: 117-27. [http://dx.doi.org/10.1177/027046769901900206]

[53] Budin-Ljøsne I, Harris JR. Ask not what personalized medicine can do for you--ask what you can do for personalized medicine. Public Health Genomics 2015; 18(3): 131-8.[http://dx.doi.org/10.1159/000373919] [PMID: 25766382]

[54] UNESCO. Universal Declaration on the Human Genome and Human Rights Available at: http://www.unesco.org/new/en/ social-and-human-sciences/themes/bioethics/human-genome-and-human-rights/ [Accessed on July 21, 2018]

[55] UNESCO-International Bioethics Committee. Universal Declaration on the Human Genome and Human Rights Paris 1997. Available from: http://www.unesco.org/new/en/social-and-human-sciences/themes/bioethics/human-genome-and-human-rights/ [Accessed on July 21, 2018]

[56] Ethics Committe HUGO. Statement on benefit sharing. Eubios J Asian Int Bioeth 2000; 10: 70-2.

[57] HUGO Ethics Committee. Statement on Human Genomic Databases. Eubios J Asian Int Bioeth 2003; 13: 99.

[58] McGuire AL, Oliver JM, Slashinski MJ, *et al.* To share or not to share: a randomized trial of consent for data sharing in genome research

genome research participants' perspectives. Public Health Genomics 2012; 15(2): 106-14.[http://dx.doi.org/10.1159/000334718] [PMID: 22213783]

[60] Bylstra Y, Lysaght T, Thrivikraman J, Watson S, Tan P. Ethical frameworks for obtaining informed consent in tumour profiling: an evidence-based case for Singapore. Hum Genomics 2017; 11(1): 31.[http://dx.doi.org/10.1186/s40246-017-0127-1] [PMID: 29221462]

[61] Kaye J, Whitley EA, Lund D, Morrison M, Teare H, Melham K. Dynamic consent: a patient interface for twenty-first century research networks. Eur J Hum Genet 2015; 23(2): 141-6.[http://dx.doi.org/10.1038/ejhg.2014.71] [PMID: 24801761]

[62] 1997.http://findarticles.com/p/articles/mi_m1272/is_n2625_v125/ai_19496201/

[63] Van Delden B, Kalis D. Leufkens. Tailor made pharmacotherapy: Future developments and ethical challenges in the field of pharmacogenomics. Bioetics 2004; 4: 305-19.

[64] Council of Europe, Reccomandation 1512 ,2001, on the Protection of the human genome, Strasbourg, Council of Europe

[65] Hoeyer K. The ethics of research biobanking: A critical review of the literature. Biotechnol Genet Eng Rev 2008; 25: 429-52. [http://dx.doi.org/10.5661/bger-25-429] [PMID: 21412365]

[66] Nordal S. Privacy.M Häyry 2007; 181-9.

[67] Danmarks National Biobank. The Danish Biobank Register Available from: www.biobankdenmark.dk/ [Accessed on January 21, 2019]

[68] Christensen H, Nielsen JS, Sørensen KM, Melbye M, Brandslund I. New national Biobank of The Danish Center for Strategic Research on Type 2 Diabetes (DD2). Clin Epidemiol 2012; 4: 37-42.[http://dx.doi.org/10.2147/CLEP.S33042] [PMID: 23071400]

[69] Van den Eynden J, Descamps T, Delporte E, *et al.* The genetic structure of the Belgian population. Hum Genomics 2018; 12(1): 6. [http://dx.doi.org/10.1186/s40246-018-0136-8] [PMID: 29394955]

[70] Tiller Jane, Otlowski Margaret, Lacaze Paul. Should Australia ban the use of genetic test results in life insurance? Perspective published: 13 December 2017[http://dx.doi.org/10.3389/fpubh.2017.00330]

[71] Juengst E, McGowan ML, Fishman JR, Settersten RA Jr. From "Personalized" to "precision" medicine: The ethical and social implications of rhetorical reform in genomic medicine. Hastings Cent Rep 2016; 46(5): 21-33.[http://dx.doi.org/10.1002/hast.614] [PMID: 27649826]

[72] Recommendation Rec(2006)4 of the Committee of Ministers to member states on research on biological materials of human origin Available from: https://www.coe.int/t/dg3/healthbioethic/Activities /10_Biobanks/Rec%282006%294%20EM%20E.pdf [Accessed on July 21, 2018]

[73] O'Doherty KC, Hawkins A. Structuring public engagement for effective input in policy development on human tissue biobanking. Public Health Genomics 2010; 13(4): 197-206.[http://dx.doi.org/10.1159/000279621] [PMID: 20395688]