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## Moving bioethics beyond ethics: a new role for the state?

*Towards a social and political approach to the public governance of the new  
biomedical technologies*

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### ABSTRACT

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Focusing on the issue of genetic diagnostic testing and drawing on a series of semi-structured interviews with genetists, epidemiologists and clinicians in Spain, this paper highlights the limits of an individualistic approach to biomedicine, embedded in a larger process of biomedicalization of the health care system and geneticization of the medical research. In contrast to the current approaches on biomedical regulation, generally based either on bioethical considerations or on technical expertise, the present work suggests the necessity of integrating the decision making process with new approaches studying the social and political consequences of the massive implementation of biomedical technologies. Through the restoration of the centrality of the political discourse, new and effective systems of governance may allow the fertile participation of all the actors involved in the production, promotion, regulation and consumption of the new biotechnological treatments whilst, at the same time, reconcile high participation with decisional efficacy.

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# Moving bioethics beyond ethics: a new role for the state?

*Towards a social and political approach to the public governance of the new biomedical technologies*

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## PREFACE

In 2002, while half in the way of my PhD studies, I had the chance to have a phone interview with one of the main officers of the UNESCO International Bioethical Committee, whose name, for privacy reasons, I will not mention here. The interview was related to my application for a stage at the IBC. The UNESCO Officer listened to me carefully, while I was trying to explain why my research would have greatly benefited from a stage at IBC. However, at some point, the officer asked me what my research background was. At first, given my multidisciplinary background, I did not know how to answer but then I replied: I am a social scientist. Suddenly, the officer put our conversation to an end saying: “I see, but.. you know.. here at IBC we work on bioethical standard setting, we may need natural scientists, jurists and even moral philosophers but surely not social scientists. You should try at the MOST (Management of Social Transformation) Unit.”

## INTRODUCTION

Whilst there exists an ever-growing literature on the ethical and legal implications of medical biotechnologies, at present there are only few up-to-date studies on their political and social implications. However, the rapid development of the new biomedical technologies is forcing policy-makers to face new types of decisional challenges. The first challenge comes from technologies like reproductive cloning that are ethically questionable but, given their expected limited diffusion, are not likely to have a remarkable social impact (G. Saint-Paul, 2003). The second type of decisional challenges may derive from technologies, like genetic screening techniques, that meet ordinary ethical requirements but, due to their potential diffusion, may carry a very high social and political impact (Shakespeare 1998, Petersen 2006). This second type of decisional challenges is precisely what this paper focuses on.



In spite of the eminently social nature of many of the issues associated with these techniques, the debate has been so far largely framed either on techno-scientific or in ethical/legal terms. The first generally focus on technical issues, such as the delivery of professional guidelines to secure international high quality standards of biomedical drugs, whilst the second are usually confined to normative issues and individual rights discourses. Either way, the result has been often the marginalisation of the political discourse and the de-responsabilization of political actors (Bora 2005, Petersen 2006). Drawing inspiration from some pioneer studies on biomedicalisation (Clarke 2003) and geneticization (Petersen 2002, 2006), this study aims to contribute to this research field through a social and political analysis of the challenges that the diffusion of genetic screening practices increasingly poses to the current system of governance of biomedical innovation and healthcare policy.

In the first section, the paper tries to explore the extent to which some of the trends associated with *biomedicalisation* (Clark 2003) may also be affecting the European research and development agenda and the national healthcare systems. As geneticization of medical research and health care policy was identified by Adele Clarke as one of the driving forces behind biomedicalisation, in the second section the paper focuses on the relationship between the diffusion of some genetic testing practices and a) the increasing emphasis on the genetic aspects of health, disease and reproduction b) the emergence of new healthcare policies based on a mix of predictive medicine, risk assessment methods and patient choice principles. Increasingly anchored to a controversial ‘right to be healthy’, as opposed to the traditional right to have access to healthcare services, these new policies seems to be supporting i) a geneticization of the medical research agenda and ii) the diffusion of new forms of genetic discrimination, whose ultimate risk is the endorsement of so called ‘weak eugenics’ selection processes.

In the light of these suggestions, the paper explores the current situation in Spain in the third section. After having presented the main characteristics of the Spanish health care system, the paper analyses the social and political issues associated with genetic testing, focusing on their current diffusion and drawing from semi-structured interviews with nine experts, who belong to different fields of expertise but ordinarily deal with genetics. In sum, both the economic indicators and the interviews seems to confirm some of the hypotheses formulated in the literature review. More specifically,



Spain seems to be experiencing a geneticization of research agenda whilst the generalisation of pre-natal genetic testing is, in fact, endorsing the diffusion of selective reproduction and clinical abortion.

Consequently, it seems clear that the large-scale implementation of genetic technologies do not raise merely technical or ethical issues, but also socio-political ones, which the current technoscientific and/or bioethical approaches seems to be cognitively and structurally unable to address. Although some novelties may come soon from very recent initiatives, such as the PHGEN (Public Health Genomics European Network) Spanish National Task Force, the paper comes to the conclusion that current systems of health care regulation and policy have been so far poorly participative and too limited in their scopes to address the social and political challenges posed by the actual and foreseen implications of the current genetic testing practices.

Therefore, in the last section, I have been tentatively outlining some proposals to develop new forms of participatory governance, in which a) the knowledge basis may be broadened as to include new insights from empirical studies on the social and political impact of a large-scale implementation of the new biomedical technologies, b) the main institutional political actors may actively and successfully respond to the above mentioned challenges, and 3) effective participation of all the actors involved in the production, promotion, regulation and consumption of the new biotechnological treatments may be reconciled with decisional efficacy.

## 1. Biomedicalisation, healthcare and social change

Although far from having reached commercialisation, many of the new biomedical technologies have already raised expectations about health care policies, medical progress and life expectancy; have attracted huge economic investments, mobilised political interests, raised political concerns and encouraged new forms of political participation. In Italy, for instance, the prospect of endorsing the research on embryonic stem cells has provoked the emergence of new political clusters, not always overlapping with the traditional political divide between centre-left and centre-right. In addition, the advances of biomedical technology have been recently associated to the emergence of a new system of technology innovation, based on new economic actors characterised by a hybrid connection between scientific and entrepreneurial profiles (Lewontin 2000).



Adele Clarke (2003) argued that the recent technoscientific innovations in biomedicine are encouraging a transition from medicalisation to *biomedicalisation*, which is shifting the emphasis from enhanced control over external nature to the harnessing and transformation of our internal nature. In this transition, molecular biology and genetics played a crucial role, producing a *geneticization* of the research and medical approach, which now assumes that is cheaper and more effective to genetically redesign the problematic bodies rather than to treat specific problems of that body.

Moreover, with its emphasis on risk assessment and surveillance, biomedicalisation is transforming the concept of 'health' into "*an individual responsibility to be fulfilled through improved access to knowledge, self surveillance, prevention, risk assessment and the consumption of self help biomedical goods and services*" (Clarke et al. 2003: 162). Consequently, biomedicalisation is encouraging an *extension of medical jurisdiction*, in the sense that various aspects of human life previously outside medical jurisdictions come to be constructed as medical problems. Second, it is also engineering a process of *commodification of health*, in which the large investments made by the public sector are socialising the costs of innovation without socialising its profits. Third, it is encouraging a *reduction of public investments in 'ordinary' academic research* and didactic activities, which is forcing the academic system to rely more and more on the funds provided by the private foundations and corporations, re-orienting their research agenda to the issues and needs of the funding agencies.

In the following, I will try to explore how some of the dynamics associated with the transition from medicalisation to biomedicalisation may be affecting the European social and economic context in which the new medical biotechnologies are going to be commercialised. This section is mainly based on a comparative analysis of the data provided by the EU Commission, the OECD Report on Biotechnologies, the OECD Health Data 2005, the OECD Report on Biotechnology and Healthy Ageing (2002) and the WHO World Health Report 2006.

According to the EU Commission data, in 2005 biotechnology has become the most funded research sector of the European Union. More specifically, in the years 2003-2005 the EU Commission has allocated 255 millions euro to the biotechnology sector, as opposed to the 232 millions to the research programme on the structuring of the EU Area and to the 227 million allocated to nanotechnology.



The OECD report on Biotechnologies provides us with some valuable information about the market potential of biotechnologies. The biotech sector in Europe is generally characterised by small to medium size firms and the countries with the largest number of firms are France and Germany. Some European countries, such as Iceland and Denmark, have actually chosen to make biotechnology one of their main fields of development and show very high biotechnology intensity scores. Concerning the public investments in the biotech sector, Spain scores very well, followed by Finland and Denmark. In contrast, looking at the data on the employment, the leaders in Europe are Germany and the United Kingdom. The overall biotechnology sales reveals a dominant position of the US, whilst in Europe, Germany, UK and Denmark hold leading positions. In the OECD countries, health dominates biotechnology applications, with more than fifty per cent of total applications in biotechnology, more than eighty per cent of total biotech R&D expenditure, and more than sixty per cent of total sales. At the EPO, biotech patents applications grew at 8.3 % every year between 1991 and 2002 and now constitute 5.3 per cent of all patent applications. Thanks to a remarkable boost of venture capitals between 1994 and 2000, the European Union now accounts for 34.5 per cent of all biotech patent applications.

The combined analysis of the two reports seems to confirm that the biotechnology sector is becoming one of most important economic sector not only for research and development but also in terms of economic growth and commercial potential. Considering that the large majority of biotechnology resources and sales relate to the health sector, I have tried to figure out the most recent situation and trends of this sector by comparing the data made available by the WHO and OECD on health expenditure and on drug expenditure, both in the private and in the public sector.

In the OECD countries, general health expenditure, both private and public, has significantly increased between 1999 and 2003. The same trend affected the public expenditure on health per capita. In the countries where the health system is essentially public, we would expect the public budget bearing the large majority of the burden. However, the public share of the general expenditure on health has not significantly increased. These data, therefore, seem to indicate that the increase in expenditure has affected proportionately more the private sector, because of a general *increase* of out-of-pocket expenditure. As to the pre-paid plans, in the UK the expenses for pre-paid



plans as part of total private health expenditure have significantly increased but in the remaining countries they have only moderately increased. The general trend, however, confirm that both public and private expenditure, at least in absolute terms, have been significantly increasing since 2002.

In spite of all the investments and expenditures, the OECD Report on Biotechnology and Healthy Ageing (2002) warns us on the real contribution of the new biomedical technologies towards health improvement. Whilst it is true the quality and the length of life has improved in the OECD countries, the report argues that such a improvement is due much more to environmental, hygienic, nutritional and educational improvements than to the advances of medical technologies. In contrast, the speed of production and diffusion of new medical technologies seems to be accountable for more than fifty per cent of the recent growth of healthcare expenditure. The *production* of new medical technologies is encouraged by the diffusion of the new academia-industry hybrids, the rapid growth of government funds and the incentive provided by the patenting system. The *diffusion* is then encouraged by various supply-side incentives currently at work in the private and in the public health care systems, which are committed to increase the intensity of their care. On top of that, the diffusion of the new medical technologies is further encouraged by the frequent misuse of their application.

The report comes to the conclusion that the overall contribution of medical biotechnology to date has been significant in economic terms but very small in therapeutic and clinical terms. In spite of the limited clinical results, the mutual reinforcement between the enormous economic interests and the huge medical expectations around biomedicine have the potential to activate some of the biomedicalisation dynamics also in European countries. However, having different R&D and health care systems, the european countries may not necessarily experiencing similar outcomes. Across the European countries, the public budget pays on average 75 per cent of total health expenditure. Consequently, the worst scenarios evoked by the prospect of biomedicalisation seem to be nowadays realistic only for the UK, where the health care system increasinly relies substantially on private actors and private health insurance. In contrast, where the health care system is public based, the expensive dynamics of biomedicalisation will mainly affect adversely other sectors of public services and provisions, such as education, transports, public infrastructures and pension



schemes. The state will be expected to select the biomedical treatments to be included under public provision, facing dramatic political and social issues, such as the generalisation of negative eugenics practices. The increasing costs will also force a rescheduling of welfare priorities, especially with regards to pensions and public healthcare provisions. In addition, the limited amount of public resources will increase the level of political tension among the various stakeholders lobbying to have their interests rewarded.

## 2. The social and political implications of genetic screening practices

Inspired by the studies on biomedicalisation, recent scholar attention has been focusing on the relationship between the diffusion of genetic screening practices and the phenomenon of geneticization. *Geneticization* can be defined as the overwhelming emphasis on the genetic aspects of health and disease, arguably affecting medical research and clinical practice, health policy as well as current social understanding of health, disease and reproduction (Lippman 1992, 1994; Hedgecoe 1998, 2001, 2002). Geneticization is a highly problematic phenomenon because it seems to reinforce social and political discrimination on the basis of genetic characteristics, whether visible or invisible (Phelan 2005).

A recent study on the generalisation of a mass neo-natal screening for cystic fibrosis (CF) in France casts some light on the mutual constitutive relationship between biomedicalization and geneticization (Vailly 2006). On the basis of a series of semi-structured interviews with government officials, Vailly shows how the implementation of CF genetic screening as a mass medical practice, initially proposed by a small group of paediatricians and bio-molecular researchers, began to be successful when both the pharmaceutical industry and some patient groups got involved. The combination of the efforts of these three actors succeeded in winning first state support and, finally, the generalisation of the practice in the French health care system. The potential generalisation of the technique also caused a remarkable change in the way the CF patient groups framed the disease and the relative action. Until 1999 the CF associations were generally negative about genetic screening and emphasised the importance of the public programmes of support for the patients. After the constitution of an alliance with some pharmaceutical industries and some biomedical researchers in 1999, the CF associations



changed opinion and presented CF as a public health problem requiring mass public action. The new social block persuaded the policy-makers to increase public funds on this specific research and to adopt the screening test into the ordinary health care system. In turn, the genetic screening has produced a serious shift in both medical and scientific practice: first it concentrated its efforts more in the ‘prevention’ of CF than in the treatment of its symptoms and, second, it has also profoundly changed the concept of ‘prevention’. The latter, in fact, no longer refers to the conditions encouraging the development of the disease (primary prevention) but to the actual reduction of the incidence of the disease in a given population (secondary prevention).

A similar research has been conducted by Carine Vassy on the Down syndrome (2006). Vassy emphasises how biomedical researchers progressively turned into moral entrepreneurs and promoted the diffusion of generalised pre-natal genetic screening for Down syndrome. After carrying out some pioneer tests at the end of the Seventies, these researchers began constituting a small network of patients groups, professionals and pharmaceutical industries. At the end of the Eighties, the network launched a massive media campaign and successfully lobbied on public authorities. Consequently, in spite of various technical and medical controversies, the public authorities decided to incorporate the screening technique as a routine practice into the public health care system. The progressive generalisation of the practice gradually promoted general consensus on medical abortions and the socialisation of costs of the screening practices, on the ground that it is cheaper to generalise the screening than to provide assistance to the individuals affected by the Down syndrome (Wald *et al.* 1992).

From a *social* point of view, the campaign for the generalisation of the DS screening techniques has been usually presented as being conducted “in the name of women” and justified in terms of a supposedly vast social demand. The paper, however, shows clearly that the social demand is the *result* of the generalisation of the screening practice rather than the *cause*, and that women’s associations as such remained for a long time uninterested and got involved into the debate only very late. From an *ethical* point of view, the legitimacy of the screening practice has been framed and justified in terms of the individual right of self-determination and choice, usually associated with the norm of informed consent. Yet, the knowledge of the condition of the foetus produces the obligation to make a choice, which, in turn, makes parents *responsible* not only for the



birth of the child but, in a sense, also for the fact that the child carries the disease. Although parents remain free to decide whether to carry on pregnancy or not, the choice make them indirectly accountable. In other words, parents who now opt for carrying on the pregnancy may be considered socially accountable for the socio-economic burden associated with carrying the disease *and* for all the possible problems the child may experience in his/her life.

In fact, the DS screening is a clear example of how a medical problem with a strong social cause – the increasingly late age in which women give birth for the first time – has been framed entirely as a genetic problem and ‘solved’ through mass genetic screening and medical abortion. The patient associations refuse to consider these practices as eugenics on the grounds of individual autonomy and informed choice (EAGS, Final Report of the workshop on *Genetic testing: challenges for society*, 2001: 10). Yet, the informed choice argument does not exclude these practices from eugenics, which includes *negative eugenics*, i.e the reduction of genetically defective people in the population (Huxley 1964).<sup>1</sup> Although under current bioethical approaches these practices may be considered ethically acceptable, they constitute a challenge promoting social and political change in the general understanding of health, disease and reproduction, whose ultimate risk is the endorsement of selective reproduction and genetic discrimination, either involving the state or simply left to the market forces.

A boundary work on *in vitro* fertilisation and pre-implantation genetic screening may actually cast some light on this issue (Ehrich *et al.* 2006). According to this study, the UK concept of welfare is experiencing a radical shift from a social welfare to a biomedical, genetic welfare. The overwhelming emphasis of the IVF/PGD staff interviewed on the genetic aspects of human health and well-being reveals the existence of a tension between a former social view of welfare, which sought to reconstitute the environment in order to accommodate the special needs of given social groups, and a new biomedical welfare that seeks to biologically refashion the problem by selecting the prospect individuals according to the biological standards currently upheld by the society. The crucial point is that our societies will be soon forced to decide whether they want to achieve justice and equity through social actions and policies or through the breeding

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<sup>1</sup> T. Shakespeare defines these practices as ‘weak eugenics’ (1998), whilst, according to the Routledge Encyclopedia of Philosophy (1998), all the practices aiming at the improvement of the human genetic pool should be considered eugenics.



selection of genetically resistant individuals, given that the two options – considering the current budget restraints on welfare provisions – may be at odd with each other. In the biomedical view of public welfare the solutions to reproductive problems are sought through an intervention at the individual genetic level that leaves the social context unaltered. On the one hand, there is a risk that the achievements in terms of social justice and social support for people carrying disabilities may be reconsidered and, in the long run, neglected. On the other hand, there is evidence to suggest that, under the biomedical, individualistic welfare model, the western society may experience a radical redefinition of citizens rights and responsibilities.

For instance, Petersen (2002, 2006) has shown how under the new patient choice health care model, citizens in the UK are expected to take greater responsibilities for managing their own health risk. In turn, citizens have been asked to improve their general medical awareness and information, which, again, has encouraged citizens to ask for more tests, more visits, more or anticipated treatment. In this respect, the diffusion of genetic screening seems to speed up the process of biomedicalisation detected by Adele Clarke by medicalising healthy individuals into pre-patients who live under the social and psychological pressure of either ‘taking care’ of their life habits or entering early treatment, whenever possible. *“There is potential for people, who have been genetically clustered into risk categories and do not take up preventive action, to be seen as failing in their duties of citizenship”* (Petersen and Bunton 2002: 1223). The public governance of population’s health is, thus, achieved through the active participation of citizens now constituted as ‘pre-patients’, because of the genetic risk they carry, and *potential consumers*, for all the drugs and treatments they may have access to (Castiel *et al.* 2006).

Although basically confirming these trends, other empirical researches do not uphold such pessimistic views. A qualitative empirical research on the impact of adult pre-symptomatic genetic testing conducted in 2006 by Paula Sakko and other collaborators, for instance, does not seem to uphold the radical view expressed by Petersen and Bunton in 2002. Sakko’s findings, based on a series of semistructured interviews, show that, regardless of their educational level, the people did not really change their lifestyle after they had underwent thrombophilia genetic testing. Yet, their study confirms the mutually reinforcing relationship between genetic screening and



biomedicalisation: the people interviewed affirmed that they would rather prefer to begin medical treatment than to change their life-style.

A recent study on the introduction of predictive medicine in life insurance shows that lifestyle information have a comparatively stronger impact on insurance behaviours than genetic information. However, it is true that risk assessment techniques, genetic and non-genetic, has encouraged a shift in the concept of health which is no longer defined as absence of disease but as absence of an increased risk of developing a disease. In addition, the insurers do consider risk factors already as if they were diseases, transforming epidemiological risks into pre-existing disorders (Van Hoyweghen *at al.* 2006: 1229). The introduction of predictive medicine has actually produced a rise of insurance premiums, notwithstanding the fact that the general health conditions have, on average, improved over the same period. With health increasingly being associated with ‘virtuousness’, the disease begins to be framed again as a ‘punishment’ for a reproachable behaviour, almost as a right reward for a bad moral conduct. Under these assumptions, if genetic risk assessment comes to be explicitly associated with an unhealthy lifestyle, the outcome may be the rise of a discriminated, *un-insurable* cast of people.

If the insurance logic begins to be applied to public health care, the very concept of citizenship may be radically transformed. If the public health care begins sanctioning on the basis of life-style conduct and/or high risk genetic profiles, being a citizen will no longer be sufficient to get access to health care support: one will need to demonstrate that he/she is either a ‘good’ citizen or a ‘low-risk’ one. Either way, the basic concept of citizenship and citizen’s rights will be dramatically transformed. The proposal made recently by Tony Blair to modify the British health care system as to adopt the first criterion seems to be the most recent step into the direction above mentioned. Apart from the issue of discrimination, Blair’s proposal is based on very controversial neoliberal assumptions about individual responsibility and health care expenditure. In fact, the rise of health expenditure is not due to the worsening of general health conditions, which have actually improved, but to the enormous uptake of medical technologies and drugs (OECD Report 2002). In addition, the patients who might be discriminated because they are smokers, for instance, have *already* paid their contributions to the health care system *twice*, that is through both the income tax and the indirect taxes on tobacco. Third, this system holds a pure individualistic approach, which entirely neglects the social factors – i.e. education,



income, family conditions – associated with disease prevalence and life habits in the population. Finally, how can the state legally approve of tobacco consumption and then penalize tobacco consumers?

The proposal advanced by Blair has to be considered within the broader debate on a on the principle of patient choice in health care that is taking place in the UK after the approval of the health care reform of 2005. On the one hand, the patient choice model attributes much more responsibility to health management to the citizens, upon whom the ultimate responsibility of preserving healthy conditions is placed. On the other hand, it claims to ‘empower’ citizens by involving them at all times from diagnosis to the final treatment and by providing them with a large amount of information and a variety of choices in terms of doctors, places and treatments. The most controversial aspect is that for the patient choice model to work, it necessary to redefine the identity of the patient. Whilst in the traditional understanding patients were not meant to choose, in the new system, extending the logic of the market to the health care system, the identity of the patient has to be constructed into a *consumer*.

The basic idea behind this transformation is that the patient-consumer, is the best judge of the value of the health care process and outcomes. Once framed as a consumer, the patient is entitled with the right/duty to choose among alternatives in an open market, which now advocates the retreat of public health care provision to give space to private actors, arguably capable of providing a largest variety of options (Propper *et al.* 2006) A broader variety of options and a free market, however, would be useless if the patient-consumers do not possess sufficient information to make choices. The increased diffusion of genetic screening seems to enter the process at this point by providing predictive information, which, through risk assessment profiles, helps to transform citizens in patients and patients into consumers. In turn, the process of empowerment makes the patient more responsible but also *more accountable* for the choices he/she makes not only in relation to the medical treatments he/she opts for but also for the lifestyle he/she adopts. This is the final twist of the patient choice revolution that has ultimately led Blair to propose sanction for people who conduct an unhealthy lifestyle.

In relation to genetics and reproduction, a recent empirical study on the actual impact and feasibility of the patient choice approach seems to radically challenge the intellectual viability of the principles of choice in health care. The study, conducted by



Skully, Banks and Shakespeare (2006), focuses on the issue of choice related to pre-natal sex selection and comes to the conclusion that very often parents do not uphold neither a technical nor a utilitarian concept of choice. Rather, they formulate their choices on the basis of ethical and existential values, according to which sex selection is not desirable as it “*is an expression of parental preference instead of a response to the future child’s needs*” (Skully *et al.* 2006: 21). The prospective parents interviewed, in fact, seemed to believe that ideal parents must be prepared to give up some of their parental control *even when such control is technically possible*. This argument runs explicitly against the main assumptions of current secular bioethics views, which consider pre-natal selection as increasing reproductive choice, contributing to individual happiness and enhancing autonomy. The parents interviewed, in contrast, proposed the alternative concept of ‘relational autonomy’, according to which good parents should pose a moral limit to their self-determination in order to preserve the child’s right to his/her autonomy. Within this framework, autonomy is not an “*intrinsic property held or expressed by an individual but a feature of a moral agent that comes into existence and is maintained through the agent’s interdependencies with, and responsibilities towards, the others*” (Skully *et al.* 2006: 30). Basically, the legitimate degree of choice with which parents may be endowed should be measured not against the abstract atomized background of a purely unconstrained individual but in relation to the degree of autonomy of the prospect child, because the parents’ autonomy only exists and makes sense within their relationship with the child.

With its emphasis on the genetic aspects of these issues, the combination between biomedicalisation and genetisation – although possibly not as radically as often argued – seems to be encouraging new forms of genetic reductionism, emphasising human biological characteristics at the expenses of social and cultural ones, but also neglecting the mutually adaptive relationship between the human beings and the environment (Castiel *et al.* 2006). In addition, there is evidence to suggest that the former combination, through the diffusion of genetic screening practices, has also established a mutually constitutive relationship with the patient choice narrative about individual health management and the transformation of the health care system.

Consequently, the social and political implications of the interaction among biomedicalization, geneticization and patient choice health care model may now be summarised as follow. First, they are encouraging the transformation of citizens into



patients-consumers and of the health care system from a public service to a private market, through a gradual retreat of public health care institutions and a parallel proliferation of private actors. Second, they are provoking an increasing demand of medical information and medical care. Third, they are orienting the research agenda towards a greater emphasis on the genetic roots of common diseases at the expenses of the studies on environmental and social factors. Fourth, they are stimulating the redefinition of the traditional concepts of health, reproduction and identity, with a greater emphasis on the genetic aspects of these concepts. Finally, they are engendering the formulation of new concepts of citizenship and political participation, in which genetic risk and virtuous behaviour become not only essential but also potentially discriminating factors.

### 3. Genetic testing, health expenditure and healthcare changes in Spain

Although only recently entered among the European Countries that have invested substantial resources on biotechnologies, Spain has managed to achieve significant results in the field by devoting increasingly larger resources to biotechnology R & D. Overall, in 2004 Spain had 284 firms active in the biotechnology firms, which accounted for about 5 per cent of the total firms active in R&D. The latter is an excellent result, especially if we consider the overall score of other European countries of similar or even bigger size. In general, Spain has been investing more in both medical and agricultural biotechnologies. In fact, Spain has one of the largest cultivations of BT maize in Europe, a result positively correlated to a low degree of politicization (PABE Report 2001) and to a relatively high level of trust that Spanish citizens show towards science and technology in general and biotechnology in particular (Lujan and Todt 2007).

However, more than half of the total investments, employment and companies in Spain are concentrated in the health sector. From an economic point of view, health biotechnology applications account on average for 66% of total sales in the biotechnology sector. Between 1991 and 2002, the percentage of biotechnology applications as a percentage of the national total of EPO doubled, rising from 2.6 to nearly 5 per cent, placing Spain more or less in the middle of the classification, following Sweden but preceding the Netherlands. The Spanish biotechnology sector is largely dependent on public actors, initiatives and funds: the public share of total biotechnology R&D amounts to 69.5 per cent, which is actually the second largest share



among the OECD countries, after Norway. The public actors involved in biotechnology also account for 69 per cent whilst private business enterprises only accounts for 33 per cent of the total. If we consider that 95 per cent of these companies have received at least some sort of public funds or contributions, the relative weight of the public sector comes dramatically to the fore (OECD Report on Biotechnology 2006).

According the WHO report on health expenditure, Spain has also been affected by a relatively high increase of health expenditures. Health expenditure per capita has increased remarkably between 2000 and 2003, going from 1038 to 1541 dollars per year, the public share of which rose from 816 to 1098. Whilst the overall health expenditure as part of the GDP has remained more or less stable around 7.5 per cent, the overall expenditure has increased on average by 2.6 per cent every year between 1998 and 2003. The increase in the private portion of health expenditure, thus, seems to be due to private pre-paid plans, which are now purchased by 1/6 of Spanish population, rather than to out-of-pocket expenses.

Although the Spanish health care system has been recently re-arranged in a strongly regionalised system, through a process of radical devolution, the most recent literature on the topic comes to the conclusion that this process has not produced substantial discrimination in access and care among the various autonomous communities (Lopez Casasnovas *et al.* 2005). Yet, depending on the system working, some regions, like Madrid, rely heavily on public institutions, whilst other regions (Catalonia, for instance) have opted for public reimbursement to private institutions. The public health care system is financed through a slightly progressive system national taxation; the revenues are later transferred to the Autonomous Communities, which spend on health between 60 and 70 per cent of their total budget. Overall, the state covers about 75 per cent of total health expenditure, whilst private expenditure accounts for the remaining 25 per cent. According to the most recent data, the increase in health expenditure in Spain confirms the suggestions made by the OECD report on Health Ageing on the impact of the large uptake of new technology and the relative abuse. In Spain, actually, ageing seems to be responsible only for nearly 8 per cent of the total health expenditure growth in 2001, whilst utilisation has been responsible for more than 33 per cent (Lopez Casasnovas *et al.* 2005: 225).



According to the IPTS report on Genetic testing in Spain, published in 2004, genetic testing has experienced a remarkable diffusion in the last decade. The large majority of genetic tests are offered and implemented by public institutions, typically hospitals, universities and research centres. Private institutions account for 20 per cent of the total number of institutions performing genetic testing. Yet, 95 per cent of all the genetic testing performed in 2001 was run on public funds. Whilst quality assurance schemes were adopted only by seven centres, informed consent was required in more than 90 per cent of the tests performed. In 2001, there were in Europe 41 genetic tests commercially available, 19 of which were commercially available in Spain. One of the most interesting findings of the IPTS report was that the frequency of the test performed was not correlated to the actual clinical value of the tests but to the dominant research trends, as demonstrated by the large number of Alzheimer tests realised, in spite of their little clinical usefulness.

Considering that one of the hypotheses derived from the literature review was that the diffusion of genetic screening techniques would encourage the large scale implementation of 'weak eugenics' measures, I will now focus for a moment on the impact of Down syndrome pre-natal screening in Spain. The Down syndrome is a genetic disorder that is associated with the presence of an extra copy of chromosome 21, and is therefore often called Trisomy. Although the causes of the mutation are unknown, the risk of having a baby carrying the genetic mutation increases with the age of the pregnant mother. The risk, however, increases slowly between 25 and 30 years old, significantly between 30 and 35, and dramatically after 35 years old (Morris *et al.* 2003). Apart from distinctive physical traits, the disease carries a series of alterations and dysfunctions, including a delay in mental development. Nonetheless, the average life expectancy of a person affected by the Down syndrome, which has significantly increased in the last decades, now approaches fifty years (Noble 1998). There are various diagnostic instruments to detect the disease, some very intrusive and risky but reliable, such as amniocentesis, and others less reliable but also less intrusive. Being the most sensitive and reliable, the genetic test is becoming the privileged practice in health care systems, as the French experience demonstrates.

In Spain, some studies have been carried out by hospitals and medical research institutions to study the prevalence of the disease (Martinez Frías *et al.* 2000; E.



Bermejo *et al.* 2006). These studies compare the data on Down syndrome births before and after the legalisation of abortion, occurred in 1985. Until then, the prevalence had remained stable around 14 children affected every 10.000 life births. In 1985, the average age of Spanish mothers at first pregnancy was about 25 years. If all of the women at first pregnancy in Spain were 25 years old, the expected amount of down life births, according to the most updated specialized literature, would be around 7 every 10.000. Obviously the real prevalence is higher, because the number of women aged between 30 and 35, which has a significant higher risk, contributes proportionally to produce higher final outcomes. In any event, starting with 1985, the actual prevalence of Down syndrome life births has decreased, initially slowly and then dramatically to the point that the current prevalence is indeed around 7 every 10.000 (2005). According to current average age of Spanish women at their first pregnancy, which is now around 29.33 years, the theoretical expected prevalence should be around 9.91, whilst the natural prevalence, considering the larger portion of pregnant women between 30 and 35, should be higher than the one registered in 1985. In 1996, for instance, the natural birth prevalence in the UK was estimated around 16.7 per 10.000 (Mutton *et al.* 1998). It is clear that the dramatic reduction is due to therapeutic abortion, which has increased remarkably since 1995.

In a study carried out in Santander in 1998, the natural prevalence has been actually reconstructed combining the data on Down syndrome life births (7), and the numbers of abortions carried out as a result of Down syndrome pre-natal diagnosis (17): the final result (24), which however includes potential spontaneous abortions, was much higher than the natural prevalence in 1985 (Martinez Frias *et al.* 2000: 406). If we look at the specific subgroups of women between 33 and 35 years, which today in Spain accounts for more than one third of all pregnant women, the impact of therapeutic abortion is dramatic: although the natural prevalence should be around 73 affected births out of 10000, the actual prevalence in 1998 was zero.

The remarkable reduction of the prevalence of Down syndrome within the population has been hailed as a great success in the prevention of the diseases. Without entering the moral dimension of the issue, I would simply underline that technically speaking the disease as such as not been prevented – given that the expected prevalence of the disease has actually been increasing along with the age of first pregnancy – but



the birth of individuals carrying the specific genetic mutation. This actually confirms the hypothesis associating the diffusion of genetic screening with a radical shift from primary prevention to secondary prevention as well as the hypothesis suggesting a shift from a social conception to a genetic conception of public welfare.

This shift is further illustrated by the recent Assisted Reproduction Law, approved in 2006 by the Spanish Parliament (Romeo Malanda 2006). The law is one of the most liberal act existing so far in Europe. Although it does not allow cloning for research purposes, it incorporates the new concept of pre-embryo; permits research on supernumerary embryos, whose production is no longer restricted; and encourages ova donation. In addition, it allows both pre-implantation and pre-natal screening. More specifically, pre-implantation screening is framed as a possible remedy to couples with hereditary genetic diseases, which now may use pre-implantation screening not only to avoid the implantation of an embryo carrying 'serious genetic diseases' but also to detect 'other alterations' that may negatively affect the 'viability' of the pre-embryo and even to select the embryo that looks more compatible with a third person (typically for transplantation reasons). The ultimate aim, as specified in the Act, is to carry on a pregnancy with 'genetically healthy embryos'.

Although implicitly referring to the hereditary genetic diseases of the parents, future extension of the 'genetically healthy' definition may become in the near future one of the most controversial concepts ever to influence our current understanding of health, identity and reproduction. The point is that, beside very few monogenetic disorders caused by the alterations of genes with a high penetrance, being 'genetically healthy' does not make sense. First, it as been estimated that each of us carries around 200 faulty genes, although often they are recessive in the pair (Morrison 2005), and second, the 'unhealthy' characteristics of the genotype have always to be assessed in relation to the biological and social environment, which is not static and change over time, largely as a result of our own actions (Vineis *et al.* 2001, Porta 2003, Hunter 2005, Luch 2005, Porta 2005).

The gradual emergence of new concepts like genetically *healthy* or genetically *compatible* seems to confirm the trend towards a more genetic and individualistic approach affecting not only the directions and priorities undertaken by the biomedical and the clinical research systems but also the new healthcare policies and the juridical



framework that underpins them. A recent issue of the Spanish periodical *Expansion* (21th March 2005) provides a further example of how the overwhelming emphasis on the genetic aspects of health, identity and reproduction is affecting the social context in Spain. According to the periodical, twenty big companies in Spain have recently offered a predictive genetic screening scheme to their top managers. When interviewed, the human resource directors have justified the initiative as a way to provide new and crucial information to their managers to help them planning their personal and family life well ahead of time. In the article, the predictive reliability of the tests is assessed at 95 per cent, a figure that highly overestimates the real predictive power of any genetic pre-symptomatic test. While the test is also available to partners and children, the results are disclosed directly to the managers, who, depending on the disease for which a risk is detected, may then ask for medical consultancy internal or external to the company. According to the data of the biotech company in charge of this service, only one out of four managers has so far refused to take the test. The most intriguing issue, however, is that the large majority of these firms have asked to remain anonymous because they were afraid that the trade unions, if informed, would seek to extend this service to all workers.

### 3.1 Genetics, healthcare and participation in Spain: the interviews

To explore the phenomenon more in depth, a series of semistructured interviews has been conducted with experts in genetic medicine across various regions in Spain. The group of experts interviewed includes one bioethicist, one expert in pharmacogenetics, two epidemiologists, an expert in health technology assessment, as well as three medical genetists (two clinicians and one researcher). The main goal of these interviews was to explore the extent and the implications of the increasing diffusion of genetic testings as they were understood by the various types of experts who actually deal with genetic testing technology. The questionnaire, structured around ten open questions, focused on five main issues: a) the contribution of genetics in medical research and clinical practice b) the nature of genetic information c) the current diffusion of genetic screening techniques and d) the idea of participatory governance, with a specific reference to the role of patient groups. The complete list of questions is



available in the annex at the end of the paper. The experts were selected by using the snowball technique, identifying first one expert for each subgroup and then asking them to suggest further colleagues to interview. At the time of writing, nine out of fifteen experts have accepted to collaborate. The interviews have been recorded, transcribed, and analysed by the same researcher.

Although the experts agreed that the contribution of genetics in medical research has been so far significant, almost all of them admit that the contribution of genetic medicine in the clinical practice so far has been marginal and largely overestimated. In case only, one genetist suggested that the relatively negligible contribution to medical practice is not due to intrinsic limits of genetic research, but to the poor knowledge and to the prejudices of the majority of the colleagues who do not know enough of genetics. Those, who are more optimistic, identify pharmaco-genetics as the very field in which genetics is more likely to positively contribute to clinical practice.

Both supporters and sceptics agree that both the private and the public research agenda have been increasingly focusing on genetic aspects of human health, diseases and reproduction, switching also resources from basic to applied research. Although this new emphasis has been increasingly securing research funds to genetic research, the genetists suggest that, compared to the huge potentials, this is far from being satisfactory. In contrast, the epidemiologists complain that this has 1) sensibly reduced public funding of epidemiological studies on complex diseases, in which the genetic profile of individuals play a limited role and 2) provoked an excessive emphasis on the genetic components of complex diseases, whilst inducing medical research to neglect the variety social and environmental factors, playing a crucial role in the development of a number of diseases. In fact, Genetist 1 affirmed “*I am a member of the evaluating committee on research proposal in biomedicine, and I can tell you: a research proposal that does not include substantial research into the genetic aspects of common diseases has not a single chance to get public funding*” (Genetist 1; questions 1-2).

Among the environmental and social factors arguably neglected by current trends in medical research, the experts have stressed the huge impact of *life habits*, like eating, smoking and drinking, and *polluting agents* present in the environment (such as lead, DDT and dioxine) for which there is evidence of much more dramatic impact vis-à-vis the development of common diseases like cardiovascular disorders and diabetes. The



totality of experts, though, agree that the genetic profile of each individual does predispose differently to the development of such diseases.

Although placing different emphasis on *how much* the genetic profile predisposes to certain diseases, the experts seemed to share the opinion that the perspective often upheld by the media and the public authorities dealing with the national R&D plans, which considers the genes *responsible* for the development of main common diseases is both *scientifically flawed* and *ideologically characterised*. Scientifically flawed because recent development in genetic research do show that the genome does not function in a deterministic way, but is largely flexible and works along a complex system of interactions among genes and between the genome and the environment.

“The genome is a complex system, it does not function in a deterministic way. Therefore, it does not really predict, it only gives us information about susceptibility and predispositions, which may, or may not, facilitate the development of a given disease, depending on various non-genetic and environmental factors. In a word, the gene does not cause, it only predisposes. (Genetist specialised in congenital anomalies).

The genetic-based perspective is also ideologically characterised because it overemphasises the role of individual genetic predispositions in order to overshadow the social and the environmental factors, and to avoid social and political responsibility for the lack of research interest and political intervention:

“The genetic variation among the human population is very small, but the differences in health conditions across the globe are huge, even within the western world. The genetic variation, at least at the population level, cannot explain neither such health differences nor their historical trends”. (Epidemiologist 2)

“There is a constant exaggeration of the genetic causes in common, complex diseases. As a result, crucial medical information about the impact of environmental and social factors, like DDT or lead contamination, is not being taken into account. In fact, we could even speak of a genetic extremism, which is literally fundamentalist, given that it pretends to deal with the foundations of the human biological nature. The governments try to make people overlook completely their political responsibility towards the social causes of common diseases, and often biotechnology and molecular biology, with false hopes and genetic reductionism, actually endorse such a trend” (Expert in clinical and molecular epidemiology)

In fact, a number of experts did not deny the possibility that the genetics may yield excellent clinical results but suggested that to achieve these results should focus



more on the actual interaction between the individual genome, the genetic alterations occurred during life-time and the environmental factors, rather than on simple causal connections between the genome and the future development of complex diseases.

The relationship between the environment and the genome is very important. Here in Spain we have, for instance, the project INMA (infancia e medioambiente) that studies, among other things, the interaction between environmental polluting agents and genetic variation in children from before birth to teen-age. (Expert in preventive medicine and public health)

Modern biology is gradually reaffirming the crucial role of the environmental factors in the regulation of genetic expression. In fact, to understand better the interaction between the genome and the environment we need to integrate epigenetic and classical genetic mechanisms” (Expert in clinical and molecular epidemiology).

It is really important, in the next ten years, how we are going to understand the interaction between the genome and the environment. A better understanding of this interaction may in fact yield important results. (Expert in genetic medicine)

In contrast to common understandings of genetic information as requiring special legal protection because of its uniquely predictive nature, all the experts affirmed that genetic information is not qualitatively different from ordinary medical information. In fact, they suggest that personal non-genetic medical information is much more reliable in predicting future diseases:

“I don’t see how the genetic information could make a patient more vulnerable than his ordinary clinical history, which, in fact, provides much more information than his/her genetic material, although in case we are talking about specific monogenic diseases the genetic information is much more relevant.” (Expert in pharmaco-genetics 1).

However, they suggest treating genetic information confidentially not so much because it is highly predictive but because it is permanent and partially shared with relatives. In general, they suggest that access to genetic information should always be granted to scientists and researchers in order to ensure further medical advances, but highly restricted vis-à-vis the general public, especially entrepreneurs or insurance companies, because it can be erroneously used as a discriminating factor (Expert in preventive medicine and public health). In fact, this seems to confirm the findings of some of the studies mentioned above, which showed how insurance companies treated risk factors as if they were some sort of disease in their own right.



The access to genetic information also raised the issue of private patenting and benefits in relation to the socialisation of research costs, which was mentioned by Clarke's study on biomedicalisation. One expert in particular expressed his/her concerns about using private genetic information, initially obtained for medical reasons, to develop tests, treatments or products later protected by a patent. In their opinion, this practice represents a form of unacceptable appropriation, which, apart from shifting benefits from the public good to private actors, does not even benefit those families providing the crucial genetic information. I quote:

It is not acceptable that genetic information obtained without any cost from affected families turns into a source of commercial profit that impose to the people affected by the same disease enormous costs. When the genetic information is turned into a scientific successful research and an important gene is discovered, these results should be freely accessible by everyone. Unfortunately, it doesn't work like this: the results of the research leads to the development of a specific diagnostic test for the discovered gene, which then gives raise to commercial exploitation (Genetist specialised in congenital anomalies).

One of the hypotheses we derived from the literature review was that the diffusion of genetic medicine may be inducing the European health care systems to switch from preventive to predictive medicine, that is from the prevention of diseases to the genetic prediction of their future development. Although some agreed that medical research efforts are increasingly focusing on predictive medicine, the experts interviewed did not detect any significant shift in the ordinary medical practice. Yet, one of them pointed out how the uptake of some genetic testing into the ordinary healthcare system did not produce any beneficial outcomes, neither at the research nor at the therapeutical level.

I believe that this shift is partially taking place. We are placing too much emphasis on the genetic causes of our common diseases, and too little on the measures that we should recommend once a genetic risk is identified. It may part of the normal process, but it is true that we did look for some studies focusing on the practices recommended to women diagnosed with BRCA1 to delay or avoid the insurgence of the disease and we found very little work done, generally of a very poor quality (Genetist specialised in congenital anomalies).

The general picture coming out of the questions on participatory governance suggests that the experts would not support the idea of enlarging participation to the



extent of giving lay people decisional power, not because of the importance of technoscientific expertise, but because they acknowledge the ultimate political nature of many of the issues related to the development and commercialisation of new genetic tests. The restricted access to the regulatory process was not justified on cognitive but on political grounds, because i) patient groups have a conflict of interests and ii) they are not democratically elected. Similar observations, though, were generally not raised against business companies, in a sense that they are often perceived as part of the game, as actors whose participation, given their propulsive role, is often taken for granted.

In contrast, the majority of the experts suggested that patient groups should merely *mediate* between the scientific community, the medical personnel and the general public. Their practical expertise may be crucial to help the public to know the problem, to raise awareness and funds, to lobby for the recognition of the medical needs of their members and finally to draw the attention of researchers and healthcare administrators to specific health issues. However, one genetist suggested that patient groups may give a crucial contribution during the first stages of medical research, because their practical expertise on the daily management of given diseases.

Although encouraging public participation at the consultative level, the experts closer to pure genetics tend to adopt a deficit model approach and to reduce the number of actors with decisional power participating to the decision making arena:

I think that the health technicians and administrators, more than politicians, lawyers or bioethicists, should be actively involved in the governance mechanisms because they know well technical and budget issues. Otherwise there is a risk that the proposals approved in the parliament on the initiatives of lawyers and bioethicists, who know nothing of the reality of an hospital or of a laboratory, later will not find financial support.

Although acknowledging that the ultimate decision on biomedical research should always belong to elected bodies, one expert in bioethics placed great emphasis on the binding power of the opinions expressed by the Spanish multidisciplinary committees on assisted reproduction and on human tissues research:

In the current legislature we are developing a system in which the opinions expressed by the specialised national spanish committees, have a binding power on the political decision-makers, in the sense that if the committee opinion is favorable to the development of a research project, the political authority may still accept or reject, but if the opinion is negative the research project cannot be latter approved by the political authority.



Only the experts closer to an epidemiological approach approved of larger public participation at all levels, including the regulatory one. They insist that an overemphasis on genetic aspects of health is excessively individualizing medicine, neglecting the social and the environmental factors, which affects us all and which, therefore, should be considered and addressed by as many actors as possible.

Finally, the expert on health technology assessment mentioned the recent implementation of a network of national committees on Public Health Genomics (PHG), as an attempt to constitute new participatory models of governance. The PhGen network, is expected to work as an ‘early detection unit’ for horizon scanning, fact finding, and monitoring of the integration of genome-based knowledge into public health (<http://www.phgen.nrw.de/typo3/index.php>). When studying its composition, one realizes that the PhGen network involves industry representatives but leaves out patient associations and consumer groups. Yet, it suggests the involvement of social scientists on the grounds that they bring in an expertise different from the one usually provided by lawyers or philosophers. In the Spanish committee, there are two social scientists involved as such, but one has a philosophical background and the other a juridical one.

The final question on patient choice and the possibility of sanctioning the citizens who conduct an unhealthy life-style, yielded substantially similar responses, which approve of a more active participation of patients in their health management, but clearly rejects the idea of discriminating them for their lifestyle. Yet, one expert in genetics suggests that a healthy lifestyle should be considered part of the therapy and therefore should be treated like an ordinary treatment. The expert in genetic anomalies pointed out that the idea of a patient choice is currently more an ideological fiction than a reality, given that serious educational programmes have not yet been implemented. The expert on health technology assessment suggested that the emphasis on patient choice may over emphasise individual responsibility, overlooking the social factors responsible for both the development of common diseases and the adoption of unhealthy lifestyles.

In sum, both the economic indicators and the interviews seems to confirm some of the hypotheses formulated in the literature review. More specifically, Spain seems to be experiencing a geneticization of research agenda, but not of the medical practice, with the notable exception of pre-natal and pre-implantation screening. Actually, the



fears about individual genetic discrimination resulting from adult pre-symptomatic genetic testing seems to be far from being empirically justified, although the geneticization of the research agenda may give rise to a social, indirect, form of genetic discrimination. The overwhelming investments on genetic research are reducing the efforts on other types of medical research and, possibly, also on the social welfare measures to support people already affected by both common and monogenetic diseases.

The hypothesis suggesting that genetic information is not clinically but socially exceptional is also confirmed. The peculiarity of genetic data does not depend on its real predictive power, but on its permanent and shared nature and on the social perception and use by third parties. The hypothesis suggesting that pre-implantation and pre-natal genetic screening may endorse weak eugenics processes, i.e selective reproduction and abortion, is also fully confirmed. This phenomenon is also changing our conception of parenthood and reproduction, which now entails a sort of right to have ‘genetically healthy’ children. The overemphasis on genetic aspects of common diseases is reinforcing an individualistic approach to health, disease and healthcare, giving rise to a more consumerist approach to healthcare provisions, although this has not (yet) produced an over-responsabilization of the individual, which is the basis for discriminating healthcare policies.

Finally, although generally reluctant to extend decisional power to civil society actors, the experts approve of general public participation and seem to be aware of the ultimate political nature of the governance system. Consequently, they know very well that the crucial issue is not the decision-making but the agenda setting processes, where they lobby and struggle exactly like any other social group. At this level, depending on their own agenda priorities, some experts want to include civil society organisations whilst others are ready to accept only the presence of industry representatives.

#### 4. Moving bioethics beyond ethics: a new role for the state?

As we have seen, human genetics poses novel regulatory problems with regards to both research and healthcare issues to national policy makers, who are required to reconcile the conflicting political demands of civil society, science and industry. Given the complexity of the issues at stake and the variety of the actors potentially involved, the recent literature has predominantly focused on the formulation of new participatory



governance models and on democratisation of expertise. This literature took its start from the crisis of a policy making system based on the so called deficit model. According to the latter, the growing resistances shown by the general public to the development and the implementation of the new biotechnologies were essentially due to lack of adequate knowledge, scientific illiteracy and mistrust. Consequently, the first proposals to overcome what was essentially framed as a communicational impasse focused on improving scientific communication, reducing scientific illiteracy and building trust. This approach gave rise to a series of attempts to promote 'educational participation' (Bora 2004).

Whilst successful in improving scientific literacy and awareness, these attempts in general failed to increase trust and encourage support. They also highlighted an educational paradox, which showed how higher education and above average scientific literacy were not necessarily associated to higher support or trust. Various studies, in fact, revealed that the resistance proceeding from the public was due more to social and cultural reasons than to scientific illiteracy, lack of knowledge and education or negative mass-media communications (Luján and Todt 2007).

In the field of biomedicine and reproductive technology, the initial reaction to the failure of the deficit model encouraged the emergence of a new discipline known as bioethics (Salter and Jones 2002). According to UNESCO, which was the first institution to set up a bioethical committee (IBC), bioethics is "*a framework of thinking that relates to the principle that must guide human action in the face of the challenges raised by biology and genetics*" (UNESCO 1997). Following the example of the UNESCO IBC, several national government set up national bioethics committees to address the ethical issues associated with genetic technologies and to enlarge participation to actors external to the techno-scientific circles. To ensure neutrality, multidisciplinary and pluralism, these committees are meant to be composed of independent experts coming from a variety of disciplines.

The bioethical debate focuses merely on the ethical implications of the new biotechnologies and aims at deriving legal guidelines from 'scientific facts' and abstract universal moral norms (Evans 2000; Bora 2005). Within the bioethical debate have so far emerged two main approaches. The first and most common approach focuses on the elaboration of universal bioethical guidelines, usually based on a common secular and



humanist framework (UNESCO, Universal Declaration on Bioethics, 2005). The second one, namely *liberal eugenics*, holds a laissez-faire approach and suggests reducing the political control over the development and the implementation of the new biotechnologies in order to let the market regulate the future directions of research and innovation progress (Buchanan *et al.* 2003; Agar 2004).

After some ten years of workings, these committees have revealed a number of serious shortcomings. Whilst they proved only partially successful in enlarging participation (Bora 2004), their workings was often affected by serious impasses on some ethical issues, which bring in existential, non-negotiable beliefs. To overcome such impasses, mainstream bioethics has increasingly narrowed down the debate to very limited issues, related to privacy, individual consensus, right to objection or property rights diverting attention from the issues of politics and power, the political economy of genetic science (Evans 2002) and legitimizing specific conceptions of health, illness and healthcare, based on a peculiar understanding of individuality (Petersen 2006). Overlooking the constraints of socio-economic background, the bioethical debate frames the individual as a universal, abstract consumer capable of making free informed choices about healthcare issues. In this way, the ethical gaze focuses exclusively on the needs and responsibilities of the individual, removing social issues and social dynamics from sight. The diffusion of genetic screening technologies is positively correlated with the growing consensus on the neoliberal patient-choice healthcare model, and favours a very consumerist approach to medicine and healthcare (Henderson and Petersen 2002), which reinforces genetic reductionism (McAfee 2003).

Although it is certainly necessary to ascertain the ethically viability of new genetic technologies, ethics cannot constitute the ultimate basis for the decision making process (De Vries and Subedi 1998). When the ethical viability of a genetic technology is ascertained, its social desirability is automatically taken for granted without even trying to assess its social and political implications. Actually, the trends highlighted by the interviews and the general overview on the budget indicators seem to confirm the validity of the critiques advanced against both the technocratic and the bioethical models, which have been structurally unable to address the emerging social and political consequences, associated with the diffusion of genetic practices. Although the clinical practice has been so far almost unaffected, the research agenda seems to be increasingly being shaped by



genetics research up to an extent that seems to have no clinical justification so far. In turn, in spite of their overwhelming impact in the development of common diseases, environmental, social and cultural factors are increasingly being neglected by the research agenda. The predictive power of genetic screening techniques seems to have been largely overestimated, paving the way to a gradual diffusion of genetic tests that has not yielded significant clinical outcomes, has encouraged the genetisation of several common diseases and produced a massive recourse also in Spain to so called ‘weak eugenics’ practices, such as therapeutic abortion for selective reproduction.

The complexity of these issues and the substantial disagreement among the experts on the real contribution of genetics, on the predictive power of genetic information and on the clinical validity of genetic testing for common diseases also provide serious arguments against even the *technical feasibility* of a regulatory decision-making process based merely on technocratic expertise. When dealing with governance issues, the experts have manifestly clustered around their own legitimate interests, defending freedom of research and often refusing the idea of extend decisional power to patient groups or civil society organisations. In other words, not only the experts are not able to reach a level of scientific consensus sufficient to constitute a reliable technical basis, but they are also far from being independent or objective about the main issues at stake. These findings cast some doubts about the overall contribution that technocratic expertise can provide. Whilst it may be useful and effective in addressing technical safety and quality of genetic testing (OECD Guidelines for Quality Assurance in Genetic Testing 2007), it seems to be far from being useful in terms of evaluating the prospect social – and often even the clinical – benefits of genetic screening technologies.

The outcomes of the interviews also show how the bioethical individualistic framework has become interiorised by the majority of the experts. The experts closer to a pure genetic approach framed the diffusion of genetic testing merely in terms of individual consensus, respect for individual autonomy, protection of individual and family privacy and defended it in the name of the ‘right to know’. In this respect, it is noteworthy that, although starting from different perspectives, both bioethicists and the genetists reached similar conclusions about the access to genetic testing and the model of governance to be adopted. This finding seems to confirm the idea that genetics and



bioethics mutually reinforce each other on the basis of a shared individualistic approach to respectively medical and ethical/legal problems.

In 2005, Bora outlined the structural and cognitive limits of bioethical committees, constrained into the iron cage of a technoscientific normativity in which law contributes to scientific knowledge and science to normative knowledge, confining politics to a parasite role (Bora 2005). Very recently, A. Petersen argued: “*The limitations of bioethics in addressing the unique challenges posed by genetics are becoming increasingly evident. In particular there is need to broaden the debate about the diverse implications of proposed and existing developments*” (Petersen 2006: 495). In the light of the interviews, broadening the debate may actually constitute a first decisive step to widen the focus of the debates, to enlarge participation, to restore to sight the social and political issues overlooked by the narrow bioethical perspectives. In this respect, the inclusion of social and political scientists into current bioethical committees may help to include sociological expertise into the regulatory framework, whilst the inclusion of representative from civil society organizations may bring to the fore the concrete social needs of important sections of the society.

The various attempts recently experimented in Denmark, UK and other countries, however, have shown a significant escalation of social conflicts among the various stake-holders (Bora 2007). Therefore, I wonder whether the problem would be more connected to the *quality* of the process rather than to the *quantity* of participation. In other words, the problem should be rephrased not so much in terms of how much participation is achieved but *when* and on *what grounds* is participation established.

Very often the public is asked to intervene only at the regulatory stage, when the large majority of issues and topics have been already framed and settled. This procedure does not encourage participation and often fuels on frustrations, especially because of the very asymmetric cognitive and financial position of lay public compared to the companies.. The participation of CSOs and lay public should be more profitably and effectively included more upstream, at the agenda setting stage, when the society is still considering priorities and directions, interests and societal relevance of the various scientific innovation processes.

The problems, however, do not only derives from the *moment* in which participation is invoked but also from the *intellectual framework* adopted so far.



According to Bora (2006, 2007), the crucial problem is due to the structural coupling of science and law in producing technoscientific norms. Bora argues that science and law have joined each other in a structural coupling and monopolised the debate, through the mechanism of double mis-reading. Mis-reading is the mechanism that allows structural coupling to work. In this case, external knowledge coming from societal actors belonging to societal systems other than science and law have been interpreted as a source of irritation leading the couple (science-law) to produce internal knowledge for decision-making. In turn, the synergy between science, officially providing expertise, and law, officially turning this expertise in normative decisions, has produced a technoscientific normative system that rigidly defines the legitimate objects of discussion, the specific definition of risk, carries the legitimate images of the “others” and of “society” congruent with the scientific view. Consequently, legal norms can only be criticised through legal rationality and scientific norms through scientific rationality. In other words, the structural coupling of science and law has rigidly structured the context and the rules of participatory science, ruling out non congruent knowledge, discourses and images belonging to other societal systems, especially to the social political realm. Outside these rigid structures and rules, participation simply is neither allowed nor considered legitimate. When a third party, who does not share a technoscientific normativity, comes to the fore, it is prevented from effective and profitable participation. In turn, frustration is generated and the escalation of conflict begins. Ultimately, exclusive dynamics are produced and the outcome (exclusion) is diametrically opposite to the intention (participation).

In his conclusive remarks, Bora suggests that as long as the participatory governance models endorse a normative/legalistic framing, political discourses will be excluded. As a consequence, they might not be the appropriate setting for democratic political participation. He suggests to split the settings for governance in three systems, each of which is expected to rely on a specific societal form of knowledge, leaving to the political institutions (democratically elected and therefore accountable) the main tasks of mediation, coordination, synthesis and final decisions. The three settings should be a) Participatory Technology Assessments setting (science), b) local processes of consultation with general public for broad political participation (politics) and c) legal administrative procedures (law).



Building on Bora's model, I suggest to separate the regulatory process in four consecutive steps. In the first step, the technical feasibility and safety of a given technology should be assessed. In the second step, the ethical viability should be ascertained, although not only in relation to universal ethical norms but also in relation to the specific national cultural norms. In the third step, the social desirability should eventually be addressed. At this stage the contribution of economics, social and political science to identify and assess the economic, social and political implications of a large-scale diffusion of new medical technologies may provide crucial information to help the final proposal reach the fourth stage, identified by Bora in the local processes of consultation with general public for broad political participation. In this final step, all the main stakeholders should be involved, from patient associations and consumer groups to industry representatives, from local authorities to scientific communities.

Yet, political participation comes at a cost. There can be no representation without taxation because sharing benefits imply also sharing duties. All the stakeholders participating should be asked, each in their own way, to help policy-makers to maximise the positive outcomes deriving from the introduction of a given technology and minimise the adverse social consequences. To give an example, if the generalisation of pre-natal genetic screening results in a reduction of public funds destined to social and medical support to people affected by the same disease for which the screening is introduced, the pharmaceutical industries may be asked to contribute to maintain the level of public funds constant over time, whilst patient associations may contribute intensifying their efforts toward the sensibilisation of the general public and to campaign for fund-raising.

If the principle of shared social responsibility becomes accepted, starting from the outcomes of the first three steps, the debate may truly restore the political discourse on social priorities, collective solidarity, bargaining and pluralism back to the center of the stage. On the one hand, the separation of technical committees in different procedural steps reduces the political power of technoscientific expertise without renouncing to its valuable contribution; and allows the ultimate decision to remain in the hands of the political body, granting to the State a new and central role. On the other hand, the progressive involvement of all the stakeholders, not merely on ethical or technical but also on social and political grounds, may help reconciling high participation with decisional efficacy.



## Conclusion

Drawing inspiration from some pioneer studies on biomedicalisation (A. Clarke 2003) and geneticization (A. Petersen 2002, 2006), this paper has conducted a social and political analysis of the challenges that the diffusion of genetic screening practices increasingly poses to the current system of governance of biomedical innovation and healthcare policy. After having explored the extent to which the dynamics associated with biomedicalisation may actually be affecting the European social and political context, the paper has explored the most recent literature on the social and political implications associated with the diffusion of some genetic testing practices. Drawing from a series of indicators on research and health expenditure in Spain and on a series of semi-structured interviews to Spanish experts, the diffusion of some genetic testing technologies has been identified as a crucial component of the complex interaction between biomedicalisation and genetisation, which is actually fueling on the rapid increase of health expenditure while encouraging the geneticization of the research agenda and the emergence of new health care policies, based on an individualistic and consumeristic conception of health, disease and healthcare. Although we haven't found any evidence of direct individual genetic discrimination associated with adult pre-symptomatic testing, the possibility that a wrong social perception and use of genetic information may actually give rise to discrimination in insurance and working contexts cannot entirely be ruled out. In contrast, the diffusion of pre-natal and pre-implantation genetic testing is actually endorsing so called 'weak eugenics' processes, based on selective reproduction and medical abortion, which are, in turn, shifting the emphasis of public health schemes from primary to secondary prevention.

Although some novelties may come soon from very recent initiatives, such as the PHGEN (Public Health Genomics European Network), the paper comes to the conclusion that current systems of genetic technology regulation and policy – generally focusing on bioethical or technical consideration – are poorly participative and structurally unable to address the social and political challenges posed by the actual and foreseen implications of the current genetic testing practices.

To allow the state to actively and successfully respond to the above mentioned challenges, the paper suggests to broaden the dominant cognitive approach integrating



technical and ethical considerations with new insights from empirical studies on the social and political impact of a large-scale implementation of the new biomedical technologies. The paper also suggests to separate the assessment of the new technologies in four different steps (technical feasibility, ethical viability, social impact and desirability, political consensus) carried out by different bodies. In the fourth step, on the basis of the principle of shared social responsibility, the paper suggests to involve all the stakeholders not on ethical and technical but on social and political grounds. Reducing the political power of technical expertise without renouncing to its valuable contribution, and addressing explicitly social and political issues in health care decision making processes, these proposals may help reconciling high participation with decisional efficacy.



## Bibliography

- Agar N. (2004), *Liberal Eugenics: in defence of a human enhancement*, Maldon Mass: Oxford Blackwell.
- Bermejo E. et al. (2006). *Vigilancia epidemiológica de anomalías congénitas en España: Treinta años de existencia del ECEMC*. In Boletín del ECEMC, Serie V, No. 5, pp. 57-83
- Bora A., G. Abels (2005), *Prime Report on Public participation, Stake Holders and Expertise*, IWT - Bielfeld University.  
Available at: <http://www.uni-bielefeld.de/iwt/bora/home/start/publications.html>
- Bora A. (2005) *Biopolitics, citizenship and the iron cage of law*, Discussion Paper, workshop 'Normativities', Bielfeld July 7-9 2005
- Bora A. and H. Heiko (2006). *Participatory science governance revisited: normative expectations versus empirical evidence*. In Science and Public Policy 33, 7, 478-488
- Bucchi M. (2006). *Scegliere il mondo che vogliamo*. Bologna: Il Mulino 2006
- Buchanan A., W. Brock, N. Daniels and D. Wikler (2000). *From Chance to Choice: Genetics and Justice*. Cambridge University Press: New York
- Clarke A. Et Al. (2003), *Biomedicalisation : technoscientific Transformations of Health, Illness and US biomedicine*. In American Sociological Review, Vol. 68 (2): 161-194
- Castiel L. D. et al. (2006). *Genomic risk and personal responsibility in health*. In Revista Panamericana de Salud Publica, vol. 19, no. 3, pp. 189-197+
- Conrad P. (2005). *The Shifting Engines of Medicalization*. In Journal of Health and Social Behavior, vol. 46 (March), pp. 3-14
- Craig E. (1998). *Routledge Encyclopedia of Philosophy*. London: Routledge
- De Vries R. and Subedi J. (1998). *Bioethics and Society: Constructing the ethical enterprise*. Upper Saddle River, NJ: Prentice Hall
- EAGS (2001). Final Report of the workshop on *Genetic testing: challenges for society*, 24 September 2001, Brussels
- Ehrich K. et al. (2006). *Social welfare, genetic welfare? Boundary-work in the IVF/PGD clinic*. In Social Sciences and Medicine, Vol.63 (5), pp. 1213-24
- Evans J., (2000). *A sociological account on the growth of principlism*. Hastings Centre Report, September-October, 31-8
- Evans J. (2002). *Playing God? Human Genetic and the razionalization of public bioethical debate*. Chicago: University of Chicago Press.
- Gaudillière Jean-Paul and Hans-Jörg Rheinberger (2004). *From molecular genetics to genomics: the mapping cultures of twentieth-century genetics*. London: Routledge



- GeneWatch (2004). *Human Genetic Testing and the influence of pharmaceutical industry*, parliamentary briefing No. 4, September 2004 (Genewatch website)
- Hedgecoe A. (1998). *Geneticization, medicalisation and polemics*. In Medicine, Healthcare and Philosophy, 1, pp. 235–243
- Hedgecoe A. (2001). *Schizophrenia and the narratives of enlightened geneticization*. In Social Studies of Science, Vol. 31, No. 6, pp. 875-911
- Hedgecoe A. (2002). *Reinventing diabetes: classification, division and the geneticization of disease*. In New Genetics & Society, Vol. 21, No. 1, pp. 7-27(21)
- Henderson S. and A . Petersen (2002) Eds. *Consuming Health: the commodification of health care*. London: Routledge
- Hunter D. J. (2005). *Gene-Environment interaction in human diseases*. In Nature Reviews, vol. 6, April 2005, pp. 287-298
- Lewontin R. (2000). *It ain't necessarily so – The Dream of Human Genome and other illusions*. London: Granta Books, 2000
- Lippman, A. (1994), 'The genetic construction of prenatal testing: Choice, consent or conformity for women?' In: K.H. Rothenberg and E.J. Thomson (eds.), *Women and Prenatal Testing: Facing the Challenges of Genetic Testing*. Columbus: Ohio State University, pp. 9–34.
- Lippman, A. (1992), *Led (astray) by genetic maps: The cartography of the human genome and health care*. In Social Science Medicine, Vol. 35, No. 12, pp. 1469–1476.
- Lopez Casasnovas G. et al. (2005), *Diversity and regional inequalities: assessing the outcomes of the Spanish 'system of health care services*. In Health Economics, 14: 221-235.
- Luch A. (2005). Nature-Nurture – Lessons from chemical carcinogenesis. In Nature Reviews, Vol. 5, February 2005, pp. 113-125
- Lujan J.L. and Todt O. (2007). *Precaution in the public: the social perception of the role of science and values in policy making*. In Public Understanding of Science, Vol. 16, No. 1, pp. 97-110
- Martinez Frías M. L. et al. (2000). *Significado de la cifras de frecuencia de defectos del tubo neuronal y de síndrome de Down en recién nacidos, corregidas y no corregidas por las interrupciones de la gestación tras el diagnóstico prenatal de esos defectos congénitos*. In Progresos de Obstetricia y Ginecología, 43, pp. 403-409
- McAfee K. (2003) *Neoliberalism at the molecular scale. Economic and genetic reductionism in biotechnology battles*. In Geoforum, n. 34, pp. 203-219
- Morandi Sabina (2004). *Test Genetici: che fine ha fatto la scienza?* Paper presented to the first International Congress of the Consiglio dei Diritti Genetici – Scienza e Società, 16<sup>th</sup> – 17<sup>th</sup> October 2004 Firenze
- Morris J. K. et al. (2003). *Comparison of models of maternal age-specific risk for Down Syndrome live births*. In Prenatal Diagnosis, vol. 23, pp. 252-258



- Morrison P. (2005). *Insurance, unfair discrimination and genetic testing*. In *The Lancet*, vol. 366, September 10, pp. 877-880.
- Noble J. (1998). *Natural History of Down's syndrome: a brief review for those involved in antenatal screening*. In *Journal of Medical Screening*, No. 5, pp. 172-177
- Petersen A. and R. Bunton (2002). *The New Genetics and the Public's Health*. London: Routledge
- Petersen A. (2006). *The Genetic Conception of Health: is it as radical as claimed?* In *Health: An interdisciplinary Journal for the Study of Health, Illness and Medicine*, Vol. 10, No. 4, pp. 481-500
- Phelan Jo C. (2005). *Geneticization of Deviant Behavior and Consequences of Stigma: The Case of Mental Illness*. In *Journal of Health and Social Behavior*, vol. 46 (December), pp. 307-322
- Porta M. (2003). *The Genome sequence is a jazz score*. In *International Journal of Epidemiology*, Vol. 32, No. 1, pp. 29-31
- Porta M. (2005). *La acumulación de alteraciones genéticas y epigenéticas: un proceso causal clave entre el medio ambiente y las enfermedades de etiología compleja*. In *Gaceta Sanitaria*, Vol. 19, No. 4, pp. 273-276
- Propper Carol et al. (2006). *Extending Choice in English Health Care: The implications of economic evidence*. In *Journal of Social Policy*, Vol. 35, No. 4, pp. 537-557
- Romeo Malanda S. (2006). *Luces y sombras de la nueva ley sobre técnicas de reproducción asistida*. In *Perspectivas en derecho y genoma humano*, No. 8, pp. 1-2
- Rothman B. K. (2002). *The Book of Life – a personal and ethical guide to race, normality and the implications of the Human Genome Project*. New York: Beacon Press
- Royal Society (2005), *Personalised Medicine: Hopes and Realities*, London: The Royal Society
- Saint-Paul G. (2003). *Economic aspects of human cloning and reprognetics*. In *Economic Policy*, April, pp. 73-122
- Sakko P. et al. (2006). *Are genetic tests exceptional? Lesson from a qualitative study on thrombophilia*. In *Social Sciences and Medicine*, vol. 63, pp. 1947-1959
- Salter B. and Jones M. (2002). *Regulating human genetics: the changing politics of biotechnology governance in the European Union*. In *Health, Risk and Society*, Vol. 4, No. 3, pp. 325-340.
- Shakespeare T. (1998). *Choices and Rights: Eugenics, genetics and disability equality*. In *Disability and Society*, Vol. 13. No. 5, pp. 665-681
- Skinner D. (2006). *Racialized Futures: Biologism and the Changing Politics of Identity*. In *Social Studies of Science*, Vol. 36, No. 3, pp. 459-488
- Skully, J. L., S. Banks and T.W Shakespeare (2006). *Chance, choice and control: Lay debate on prenatal sex selection*. In *Social sciences and medicine*, Vol. 63, pp. 21-31



- UNESCO (1997). *Resolution on the implementation of the Universal Declaration on the Human Genome and Human Rights*. Resolution 29 C/17, 11<sup>th</sup> November. Paris: UNESCO
- Van Hoyweghen I. et al. (2006). *Making the normal deviant: the introduction of predictive medicine in life insurance*. In Social Sciences and Medicine, vol. 63, pp. 1225-1235
- Vailly J. (2006). *Genetic screening as a technique of government: The case of neonatal screening for cystic fibrosis in France*. In Social Sciences and Medicine, vol. 63, pp. 3092-3101
- Vassy C. (2006) *From genetic innovation to mass health programmes: the diffusion of Down's syndrome prenatal screening and diagnostic techniques in France*. In Social Sciences and Medicine, vol. 63, pp. 2041-2051
- Vineis P. et al. (2001). *Misconceptions about the use of genetic testing in populations*. In The Lancet, vol. 357, March 3, pp. 709-712
- Wald N. et al.(1992). *Antenatal maternal screening for Down Syndrome: results of a demonstration project*. In British Medical Journal, 305, pp. 391-394

## Databases

- EU Commission Budget 2003-2005, European Union Website
- OECD Health Report 2006, OECD Website
- OECD Biotechnology Report 2005, OECD Website
- OECD Biotechnology and Healthy Ageing Report 2002
- OECD Health Database 2006, OECD Website
- WHO Health Report 2006, WHO Website
- UNESCO, *IBC Proceedings* 1997-2006, Paris: UNESCO Press 1997-2006
- IPTS 2002. Genetic Testing Services for Hereditary Diseases in Spain. European Commission Joint Research Centre.
- OECD Guidelines for quality assurance in molecular genetic testing, forthcoming 2007

## ANNEX I

## Questionnaire for semistructured interviews with experts in Spain

- 1) Cuál es *actualmente* la contribución de la genética en
  - ¿La investigación científica médica?
  - ¿La práctica clínica?
  - ¿El sistema sanitario en general?
- 2) ¿Cuál será, en su opinión, la contribución de la genética *a medio plazo*, o sea en 10-20 años?
- 3) ¿Qué características tiene la información genética que la hacen diferenciarse de otros tipos de información médica
  - ¿Con respecto a su fiabilidad y capacidad de predecir?
  - ¿Con respecto a su confidencialidad y su acceso?
- 4) ¿Qué factores, a parte de la información genética, pueden tener influencia en la relación genotipo-fenotipo, especialmente con respecto al desarrollo de enfermedades? y ¿cómo valoraría la importancia de la correlación genotipo-fenotipo?
- 5) ¿Conoce usted algunos estudios, proyectos u otros tipos de investigación, en España o en Europa, que estudian la relación entre genotipos, fenotipos y otros tipos de factores en relación al desarrollo de enfermedades?
- 6) En gran parte de la literatura reciente, sobre todo en el mundo anglosajón, se habla muy frecuentemente de la emergencia de un nuevo enfoque en la medicina, tanto en la investigación como en la práctica clínica, más enfocado sobre las causas genéticas de las enfermedades que en el tratamiento de sus consecuencias. ¿Cree usted que exista este cambio y, en el caso de que exista, que opinión tiene sobre esto?
- 7) En los últimos años en España, así como en toda Europa, las pruebas genéticas de diagnóstico y pre-sintomáticas, prenatal y para adultos, han tenido una gran difusión. Esta difusión ha sido defendida y apoyada por su capacidad de reducir la incidencia y el impacto de enfermedades debidas, al menos en parte a predisposiciones genéticas. ¿Cuál es su opinión?
- 8) En las última décadas las asociaciones de enfermos se han interesado mucho en el desarrollo de las nuevas biotecnologías médicas. ¿En su opinión, cuál es – y cual debería ser – el papel de las asociaciones de enfermos en
  - ¿La investigación científica médica?
  - ¿La práctica clínica?
  - ¿El sistema sanitario en general?
  - ¿La gestión y la regulación de las nuevas biotecnologías médicas?



- 9) En la literatura especializada sobre la regulación, la promoción y la gestión de las nuevas biotecnologías se propone cada día más frecuentemente el desarrollo y la adopción de nuevas formas de gobernanza, más participativa. ¿Está de acuerdo? ¿Qué enfoque deberían tener estas nuevas formas de gobernanza? ¿Qué conocimiento e información debería tener quien pretende participar? ¿Y, en su opinión, quien debería participar?
  
- 10) En Inglaterra se está desarrollando una nueva forma de política sanitaria, que atribuye mucha más responsabilidad sobre al paciente, con respecto a su conducta y a su estilo de vida, pero al mismo tiempo, le ofrece más posibilidades de elección en términos de acceso a la información, tipo de tratamiento, lugar y personal médico. ¿Cuál es su opinión sobre esto? ¿Cree que el Estado debería sancionar quien lleve un estilo de vida poco saludable?