Genetic Testing, Geneticisation and Social Change: Insights from Genetic Experts in Spain

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Introduction

Over recent years, the achievements of human genetics research have triggered a transcendental change in our understanding of the human body and of a variety of diseases with a genetic basis. These achievements (...) are, in turn, leading to remarkable advances in clinical practice. This is especially true in relation to diagnostics and the prevention of an increasing number of diseases for which we now have appropriate genetic tests (...) The extraordinary sensitivity of genetic information, however, transcends the purely individual domain, and therefore its social and ethical implications require a special regulatory frame. (Andalusian Bill on the regulation of genetic testing, 2007)

The rapid progress of medical research in genetics has raised great hopes for future disease treatments and prevention, paving the way for the emergence of new medical perspectives based on predictive medicine and personalised treatments (Horst 2007; OECD 2007; Royal Society 2005). However, given the permanent nature of genetic information, the diffusion of genetic testing technologies (GTTs) has also raised a number of ethical dilemmas, not only related to the psychological impact of testing but also in relation to privacy and discrimination (Holtzman & Shapiro 1998; Sandor 1999). Since the 1990s, the ethical implications of genetic testing and screening have been extensively studied, not only in general terms (Chadwick & Wiesing 1999; Fulda & Lykens 2006) but also in relation to specific national contexts (Dierickx 1999; Hauser 1999; Hoedemakers 1999), to specific conditions (Koenig et al. 1998; Post et al. 1997), target groups (Borry et al. 2006; Clarke 1999) and types of test (Lippman 1991; Wilcksen et al. 2003).
However, the large-scale implementation of genetic testing technologies raises not only technical or ethical issues but also socio-political ones (Davison et al. 1994; Lehoux & Blume 2000; Lock et al. 2006). More specifically, it has been argued that the current diffusion of GTTs may stimulate the early medicalisation of healthy individuals (Clarke 2003; Conrad 2005), fuel a rapid increase in health expenditure, and promote the emergence of new individualistic and consumer-based healthcare policies (Petersen & Bunton 2002; Petersen 2006). Moreover, it has been argued that GTTs may encourage a geneticisation of diseases (Lippman 1991; 1992; Hedgecoe 2001; 2002), reproduction (Lebner 2000; Lippman 1993; Shakespeare 1995; 1998) and identity (Pálsson 2007; Skinner 2006; Soo-Jin Lee 2005) and further endorse discrimination in insurance and employment practices (Dodge & Christianson 2007; Goven 2008; Markel & Barclay 2007; Sedo 2007; Van Hoyweghen et al. 2006; 2007).

These studies have dealt with a number of European and North American countries but there exists no up-to-date contribution on the social and political dynamics of geneticisation and medicalisation associated with the growing diffusion of genetic testing technologies in Spain. Yet Spain now constitutes a very interesting case, having recently made massive investments in biomedical research whilst adopting a very liberal regulatory framework. The present study aims to contribute to the debate through a social and political analysis of the interaction between genetic testing technologies, biomedical research and healthcare policy in the Spanish context.

This chapter will first provide a short overview of current social studies to highlight some of the social and political dynamics most commonly associated with the diffusion of GTTs. After providing an outline of the current state of regulatory legislation, medical research and healthcare policies, I will move on to explore some of these dynamics in a series of semi-structured interviews with Spanish experts in different fields of medical genetics. The outcomes of these interviews suggest that the diffusion of genetic testing affects medical research fields and clinical practices in different ways. In addition, it emerged that experts belonging to different fields of medical genetics have different opinions with regard to the contribution of genetics to medical practice and its impact on
healthcare policy. However, the experts share the opinion that, except for reproductive medicine, the diffusion of genetic testing technologies in Spain has so far been more closely associated with a geneticisation of the research agenda than of clinical practice.

Genetic testing, biomedicalisation and geneticisation

In 2003, Adele Clarke argued that recent technoscientific innovations in biomedicine are encouraging a transition from medicalisation to biomedicalisation, shifting the emphasis from enhanced control over external nature to the harnessing and transformation of our internal, genetic nature (Clarke 2003). This process assumes that it is cheaper and more effective to redesign the problematic bodies genetically rather than to treat the specific problems of that body. As a consequence, the genetic aspects of human life and body are likely to be overemphasised, encouraging a geneticisation of both medical research and clinical practice. In this process, a crucial role is played by the information increasingly produced by genetic testing technologies.

However, geneticisation and biomedicalisation cannot be observed in all fields of medicine in the same way, to the same extent or with similar implications. In order to get a more articulated perspective on what geneticisation and biomedicalisation actually mean, it is necessary to take into account the variety of cultural, social and political factors in which medical research and clinical practice are embedded. These factors affect nations, medical disciplines and healthcare policies in remarkably different ways. Moreover, it is also necessary to differentiate by looking into different medical fields and practices in relation to different testing technologies, as they provide distinct types of information, contributing to the broader phenomena of geneticisation in different ways (see Table 1).

In fact, geneticisation is a highly contested concept. It was first introduced by Lippman, who defined it as ‘an ongoing process by which differences between individuals are reduced to their DNA codes, with most physical and behavioral diseases defined at least in part as genetic in origin. It refers as well to the process by which intervention employing genetic
technologies are adopted to manage problems of health’ (Lippman 1991, 19). Since then, some authors have tried to link geneticisation with the pre-existing debate surrounding the emergence of medicalisation (Arnasson & Hjorliefsson 2007; Clarke 2003; Lippman 1991; 1992; 1994; 1998; Scott 2006; Ten Have 2001). Other scholars, however, rejected the idea that the advancement of genetics was actually promoting relevant medical or social change (Condit & Williams 1997).

### Table 1. Genetic tests most commonly performed in Spain

<table>
<thead>
<tr>
<th></th>
<th>Pre-implantation</th>
<th>Pre-natal</th>
<th>Neonatal</th>
<th>Adult</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Diagnostic</strong></td>
<td>CH, fragile X syndrome, haemophilia spinal muscular atrophy, myotonic dystrophy 1</td>
<td>Down syndrome, neural tube defects, MS/MS tandem mass spectrometry</td>
<td>PKU, CH (nationwide); CF, DMD, GAL, haemoglobinopathies</td>
<td>(at regional level)</td>
</tr>
<tr>
<td><strong>Predictive</strong></td>
<td>Huntington disease, DMD, CF</td>
<td></td>
<td></td>
<td>Huntington disease</td>
</tr>
<tr>
<td><strong>Susceptibility</strong></td>
<td>BRCA 1 &amp; 2</td>
<td></td>
<td></td>
<td>BRCA 1 &amp; 2</td>
</tr>
<tr>
<td><strong>Carrier</strong></td>
<td></td>
<td></td>
<td></td>
<td>sickle cells</td>
</tr>
</tbody>
</table>

**Legend:**
- CF Cystic Fibrosis
- CH Congenital Hypothyroidism
- GAL Galactosaemia
- ISNS International Society for Neonatal Screening
- MS/MS Tandem Mass Spectrometry
- PKU Phenylketonuria
- DMD Duchenne Muscular Dystrophy

In his critique of geneticisation, Adam Hedgecoe (1998; 1999) suggested that social research on geneticisation should abandon the abstract and general ground of polemics, focus on specific diseases, technologies or medical fields and extend its scope to consider the wider context, seeking empirical evidence of socially relevant change (2001). For ‘geneticisation’ should really be considered primarily as ‘a heuristic tool that can help to re-focus the moral debate on the implications of new genetic knowledge towards interpersonal relations, the power of medicine, the cultural context and social constraints, rather than emphasizing issues as personal autonomy and individual rights’ (Ten Have 2001, 295). Since then, several studies have looked at the progressive geneticisation of some specific diseases, i.e. the gradual construction of these diseases into overtly genetic conditions (Hedgecoe 2001; 2002; Hall 2005; Weiner & Martin 2008). Other studies have tried to consider empirically the impact of genetic testing in the job and insurance markets (Dodge & Christianson 2007; Goven 2006; Markel & Barclay 2007; Sedo 2007; Van Hoyweghen 2007), in general healthcare policy and institutions as well as in healthcare and clinical practices. (Kerr 2005; Skully et al. 2006; Vailly 2006; 2008). These have yielded mixed results. Although they move in different directions and adopt different approaches, these studies in fact focus on a common research question: where can geneticisation be actually seen at work and where does it play a less dominant role?

In the light of these distinctions, an overview of recent case studies may help to introduce some of the social and political implications associated with different types of genetic testing. Recent studies on cystic fibrosis (CF) neonatal screening in France have shown how CF patient groups are affected by their interactions with pharmaceutical industries and biotech researchers. Patients became increasingly focused on the genetic aspects of CF, and their interest shifted to public research on genetic screening. They consequently became actively involved in a campaign to persuade policy makers to introduce neonatal CF genetic screening as a generalised medical practice (Vailly 2006). More recently, it has been acknowledged that the concept of abnormality in CF has been expanded by the neonatal screening programme so that it now includes within biological abnormality mild forms of CF which previously escaped clinical diagnosis (Vailly 2008).
It was also in France that concerns expressed by biomedical researchers acting as moral entrepreneurs led to the spread of pre-natal screening for Down syndrome (DS). Despite various technical and medical controversies, the authorities eventually introduced the screening technique as routine practice in public health care. The progressive proliferation of the practice gradually promoted a general consensus on therapeutic abortion and on the socialisation of screening costs (Vassy 2006). The increasingly widespread screening, however, has indirectly provoked remarkable social changes for the population. For instance, access to Down syndrome tests as well as the probability of pregnancy interruption following a positive result are highly correlated to education, working position and geographical provenance. As a result, Down syndrome is becoming a disease increasingly associated with those with low education, found predominantly in working class groups of non-French origins, who may then become disproportionately responsible for the care of infants born with this congenital anomaly (Khoshnood et al. 2006).

In fact, DS screening is a clear example of how a disease condition strongly associated with a social phenomenon – the increasingly late age at which women are having children – has been increasingly framed as a genetic problem and ‘solved’ through mass genetic screening and therapeutic abortion. The generalisation of prenatal diagnostic testing, thus, seems to encourage what Shakespeare termed ‘weak eugenics’, namely, ‘promoting technologies of reproductive selection via non-coercive individual choices’ (Shakespeare 1998, 670).1

Whilst the reduction of the prevalence of Down syndrome and other monogenetic disabilities or diseases has been hailed as a great success for disease prevention, the diffusion of pre-natal genetic screenings seems to endorse the emergence of a narrowly genetic approach to the management of disease and disability prevalence in the wider population. Khoury (1997), for instance, argued that prenatal genetic testing is, in fact, shifting the emphasis from primary or phenotypic prevention, namely reducing the conditions encouraging the development of the disease, to secondary or genotypic prevention, which aims to reduce the number of affected individuals in a given population.
Pre-implantation genetic testing has similar implications and also raises new ones. Ehrich et al. (2006), for instance, suggested that as pre-implantation screening is increasingly coming to be seen as a tool for prevention, our society may be experiencing a radical shift from social welfare towards biomedical, genetic welfare (Ehrich et al. 2006). The answers given by IVF / PGD staff who were interviewed on the genetic aspects of human health suggested an overwhelming consensus. They emphasised the emergence of a tension between the 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 embryos of future individuals according to biological standards currently upheld by society. As Shakespeare suggested, there is a risk that the gains made in social justice and social support for people with disabilities may come to be reconsidered and, in the long run, neglected (Shakespeare 1995).

Finally, the diffusion of susceptibility and predictive genetic tests entails a whole new set of implications, encouraging a radical redefinition of citizens’ rights and responsibilities. Petersen (2006) showed how the new patient choice healthcare model meant that citizens in the UK are expected to take greater responsibility for managing their own health risk. Whilst people are now expected to improve their general medical awareness and information, medical institutions are increasingly exposed to strong pressures to provide more tests, more visits and the most up-to-date treatments. Within this framework, the diffusion of susceptibility and predictive genetic testing only help to further medicalise 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 & Bunton 2002, 1223).

The governance of public health is thus achieved through the active participation of citizens now constituted as pre-patients – because of the genetic risk they carry – and as potential consumers – for all the drugs and treatments they may have access to (Castiel et al. 2006). Moreover, risk assessment techniques seem to encourage a new healthcare perspective, in which
health is defined as the absence of an increased risk of developing a disease and epidemiological risks are turned into pre-existing disorders (Van Hoyweghen et al. 2006, 1229). In turn, information on genetic risk increasingly conditions lifestyle choices when it comes to determining access to healthcare support, forcing the public to demonstrate their healthy behaviours and/or their low risk genetic profiles. Either way, the basic concept of citizenship and citizens’ rights is experiencing a radical transformation. In fact, there is strong evidence to suggest that, in current debates about personalised medicine (Royal Society 2005), emphasis on genetic information engages in a mutually constitutive relationship with the patient choice narrative about individual health management and the transformation of the healthcare system (NHS Report 2003).

Finally, geneticisation processes which emphasise human genetic and biological characteristics at the expense of socio-cultural ones, tend to neglect the mutually adaptive relationship between human beings and the environment (Castiel et al. 2006). With regard to the research agenda on common diseases, this induces research efforts to focus ever more on genetic factors to the detriment of social and environmental ones.

To summarise, the process of geneticisation associated with the increasing diffusion of genetic testing technologies is stimulating the re-definition of the traditional concepts of health, reproduction and identity, placing greater emphasis on the genetic aspects of these concepts. However, the process is not homogeneous and different genetic testing techniques give rise to specific outcomes, which I have summarised as follows:

1. Neonatal screening results in a stronger emphasis being placed on the genetic aspects of certain diseases and expands the notion of biological abnormality.
2. Prenatal genetic tests endorse
   – ‘weak eugenics’ trends through ‘therapeutic’ abortion,
   – the shift of medical focus from phenotypic to genotypic prevention.
3. Pre-implantation screening encourages
   – a transition from social welfare to biomedical welfare by selecting the embryos of future individuals to meet social standards instead
of adapting society to meet their potential medical needs.
—a shift in medical focus from phenotypic to genotypic prevention.

4. Susceptibility genetic testing may
—medicalise asymptomatic individuals into ‘healthy sick’.
—turn healthy individuals into patient-consumers and increase the
demand for medical information and care.
—engender the formulation of new concepts of citizenship in which
genetic risk and / or virtuous behaviour produce differential access to
healthcare support.
—support a shift in focus from therapeutics to predictive medicine.
—induce research to place greater emphasis on the genetic roots of
common diseases at the expense of social and environmental factors.

In the remaining part of this chapter, I will analyse these social and politi-
cal dynamics, generally associated with the development and intro-
duction of genetic testing practices, as they are present in the Spanish
context.

Genetic testing, normative framework
and medical practices in Spain

Compared to other European countries, Spain turned its attention to bio-
technology relatively late, yet it has already achieved fourth position with
regard to patenting and academic publishing in this field. Spanish R&D
in biotechnology has seen 350 per cent growth over the past four years.
This has been due largely to increasing public support: 96 per cent of
business companies investing in R&D in the past four years have received
public funding (OECD 2006). The most developed biotech area in Spain
is biomedicine, accounting for over half of all biotech research and repre-
senting 35 percent of all publicly funded research (Biotechnology Journal,
Focus on Spain, 2008).

Public research in Spain is mainly funded through the Ministry of
Science and Innovation according to general R&D plans approved every
four years. The annual call for applications is issued on the basis of macro-
areas associated to a specific type of diseases with each area being divided into sub-themes and priorities. A close examination of the general aims and project priorities in biomedical research from 1996 to 2007 reveals interesting shifts and changes.

A general overview reveals that in 1996, genetic-oriented research accounted for 20 per cent of projects. In 2007, this figure rose to around 32 per cent. However, it is important to note that during the years 2001–2003, research in genetics and genomics accounted for more than 60 per cent of medical research overall. Moreover, genetic and molecular studies into cancer and neurodegenerative diseases are the single most funded research themes. In terms of sub-areas, genetic studies on cancer only accounted for about 20 per cent of research in 1996, but were more than 60 per cent in 2007. In contrast, research into cardiovascular diseases and neurological diseases maintained its initial proportion, with 20 and 50 per cent of genetic studies respectively. It is interesting, however, that within the area of neurological diseases, there has emerged a well funded research field relating to genetic predispositions to addictive behaviours (source: Ministry of Science and Innovation, unpublished data). Therefore there would seem to be evidence to (cautiously) suggest that the medical research agenda in Spain has recently been characterised by a growth in genetic studies, especially since 2001.

Like other OECD countries, Spain has also been affected by a rise in health expenditure, 75 per cent of which is funded from the public budget (López Casasnovas et al. 2005; OECD 2006; WHO 2005). Within this context, the increase in the number of genetic tests performed in Spain from 2000 to 2002 is highly relevant as the greater majority of these tests were performed and funded by public institutions (Rueda & Briones 2002). For example, in 2002, tests were available for 34 different genetic conditions, although the number of genetic variations is much larger (AEGH 2002; 2005). The majority of tests performed were either diagnostic or predictive but there also existed a significant number of susceptibility and pharmacogenetic tests. Whilst the diagnostic tests were performed by all actors, private actors specialised in predictive and predisposition tests, and pharmacogenetic tests were evenly spread between the actors (Table 2; IPTS 2002). It is interesting to note that the frequency of the
tests performed was higher for tests that are perhaps interesting from a research point of view but of little use for patients (Rueda & Briones 2002). At the time, there was no regulation in Spain governing genetic testing, and so the ISCiii report observed a significant lack of common quality standards, with regard to both official accreditation and informed consent policy.

Table 2. Genetic Testing Technologies: diffusion and potential implications

<table>
<thead>
<tr>
<th>Genetic Testing Technologies</th>
<th>Level of Diffusion</th>
<th>Current Status</th>
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<tbody>
<tr>
<td>I Neonatal screening</td>
<td>Offered on the basis of family history</td>
<td></td>
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<tr>
<td>expansion of the notion of biological abnormality</td>
<td>To be assessed</td>
<td></td>
</tr>
<tr>
<td>II Prenatal genetic tests</td>
<td>Offered to all women older than 35</td>
<td>Confirmed by Down data, literature and interviews</td>
</tr>
<tr>
<td>endorsement of weak eugenics through 'therapeutic' abortion</td>
<td></td>
<td></td>
</tr>
<tr>
<td>shift from primary to secondary prevention</td>
<td>Confirmed by 2006 law and by interviews</td>
<td></td>
</tr>
<tr>
<td>III Pre-implantation screening</td>
<td>Offered on the basis of family history and risk of hereditary genetic conditions or screening for genetically compatible siblings</td>
<td>Confirmed by 2006 law</td>
</tr>
<tr>
<td>transition from social welfare to biomedical welfare</td>
<td></td>
<td></td>
</tr>
<tr>
<td>shift from primary to secondary prevention</td>
<td>Confirmed by interviews and by 2007 Andalusian law</td>
<td></td>
</tr>
</tbody>
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### IV

Susceptibility genetic testing may:

<table>
<thead>
<tr>
<th></th>
<th>Mainly offered as part of genetic research projects in public hospitals and laboratories. Not common in ordinary clinical practice.</th>
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<tbody>
<tr>
<td>medicalise asymptomatic individuals into 'healthy sick' and change their life-style.</td>
<td>To be assessed</td>
</tr>
<tr>
<td>encourage the shift towards patient-choice model and personalised medicine.</td>
<td>No shift in actual healthcare system detected, but personalised medicine recognised as a goal in 2007 national and Andalusian law</td>
</tr>
<tr>
<td>engender the formulation of new concepts of citizenship in which genetic risk and/or virtuous behaviour produce differential access to healthcare support.</td>
<td>No relevant changes registered in medical practice or legal framework</td>
</tr>
<tr>
<td>support the shift in medical focus from therapeutic to predictive medicine.</td>
<td>Suggested by 2007 Andalusian and biomedicine law. Not yet observed.</td>
</tr>
<tr>
<td>induce research to place greater emphasis on genetic roots of common diseases at the expense of social and environmental factors.</td>
<td>Confirmed by data on research agenda and by interviews</td>
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</table>
The increase in genetic testing, however, was more a result of the growing number of research programmes than an outcome of changing medical practice, as no generalised genetic screening policy has yet been introduced in Spain and the uptake of susceptibility and predictive genetic tests in the healthcare system has been negligible so far. Neonatal screening for phenylketonuria (PKU) and congenital hypothyroidism (CH) is offered nationwide, whilst galactosaemia, CF, Duchenne muscular dystrophy and haemoglobinopathies have been introduced recently in some regions, but their application is not widespread and depends on family history (Loeber 2007). The situation is slightly different for pre-natal screening, which is the most common testing technique, offered for a variety of congenital anomalies. With regard to Down syndrome, although nuchal scans are only offered in some regions, often in private institutions, amniocentesis and chorionic villus sample (CVS) are routinely performed, and are publicly funded for all pregnant women over 35. Depending on maternal age, family history and fetal anomalies detected through ordinary monitoring practices, various other common pre-natal tests are also offered and carried out, completely subsidised by all the regional healthcare systems (AEGH 2002; Godard et al. 2003; Javaher et al. 2008; Parga Soler et al. 2001).

In Spain, the impact of pre-natal screening has been remarkable. Recent studies have compared the data on Down syndrome births before and after the legalisation on abortion (Bermejo et al. 2006; Martinez Frías et al. 2000). Prior to the abortion legalisation (1985) the natural prevalence had remained stable at 14 per ten thousand births. After legalisation, as a result of pre-natal testing being offered to all pregnant women older than 35, the actual prevalence of Down syndrome live births has gradually decreased to around 7 per ten thousand births. In fact, due to the rising age of first birth, the natural expected prevalence should now be around 17 per ten thousand births (Morris et al. 2003). The abortion rate after positive pre-natal diagnosis is currently 96 per cent (Boyd et al. 2008). Actually, a recent study which examined the information which hospital professionals provide about Down syndrome before the test is carried out, revealed that the Spanish leaflet contained only very limited information, which described the syndrome in very negative terms and clearly encouraged termination of pregnancy (Hall 2007).
Before the National Assisted Reproduction Law was approved (2006), pre-implantation screening (PGD) was not regulated, except for a specific bill passed by the Andalusian Regional Autonomy in 2005. In this bill, pre-implantation screening was limited to a number of specific conditions and required a complex procedure from request to authorisation to the testing process, in which the regional bioethics committee was also involved. However, it allowed PGD not only for early-onset, untreatable diseases, like DMD, but also for late-onset ones, like Huntington disease, and for diseases with mild to severe expressions like haemophilia A and B (Andalusian Bill on Assisted Reproduction 2005, 12).

In contrast, the 2006 national law on assisted reproduction inaugurated one of the most liberal European regimes so far. It adopts the concept of pre-embryo, permits production of pre-embryos for research purposes, and encourages ova donation. It also includes IVF and PGD on the list of medical techniques subsidised by public healthcare systems. To date, however, only Andalusia and Murcia regional healthcare authorities will cover the cost of IVF and PGD. As a result, PGD is mainly performed by private IVF and genetic centres and paid privately (IPTS 2007). The law includes IVF and PGD among techniques covered by the public healthcare budget because it classifies assisted reproduction techniques no longer merely as technological remedies to human infertility but as new instruments for the prevention of disease prevalence in the population.

(…) the techniques of assisted reproduction, apart from contributing to the overcoming of sterility problems, appear especially helpful for diagnostic and research purposes (…). Pre-implantation screening opens new ways to prevent genetic diseases that currently lack treatment and for the selection of pre-embryos (…) useful to cure a sick relative. (Law on Assisted Reproduction 2006)

More specifically, pre-implantation screening is made available to select a pre-embryo free from ‘serious hereditary diseases that lack post-natal treatment’ and also to detect ‘other alterations’ that may negatively affect the ‘viability’ of the pre-embryo. In practice, although originally confined to early onset, untreatable monogenic diseases, PGD has recently been allowed for hereditary breast cancer (BRCA 1 and 2) and Huntington disease. In fact, the definition of the law leaves great room
for future expansion of the definition of genetic diseases, paving the way to the selection of what may be called ‘genetically healthy pre-embryos’ (Romeo Malanda 2006). Yet, whilst each individual carries around 200 faulty genes, most of them recessive in the pair (Morrison 2005), the ‘unhealthy’ characteristics of the genotype must always be assessed in relation to the biological and social environment, which is not static and changes over time, largely as a result of our own actions (Hunter 2005; Luch 2005; Porta 2003; Vineis et al. 2001).

Predictive, pre-symptomatic and susceptibility genetic tests in adults are currently performed within the framework of research programmes, and are offered on the basis of family history and clinical assessment. Future changes may also be triggered by the Law on Biomedical Research (2007), which paved the way for a more systematic introduction of these tests into medical practice. This law, which regulates all areas of biomedical research including genetic testing, does not make a legal distinction between genetic tests carried out for medical or research purposes. On the one hand, it ensures that all genetic tests will be offered on an equal access basis and provided by the public system. On the other hand, it allows the introduction of genetics tests ‘to identify affected, non-affected or carriers of genetic variations that may cause, or predispose an individual to develop a disease, or affect his / her response to a given treatment’. It is important to point out that the Andalusian bill (2007) implementing the national law presents genetic testing as a crucial step towards a healthcare system based on personalised medicine and recognises each patient’s ‘right to predictive medicine’.

The national law, which allows cloning for research and medical purposes, allows hospital and genetic laboratories to perform any genetic test they regard as necessary, provided that written individual consent, genetic counselling and privacy requirements are respected. Only genetic screening programmes require the permission of regional healthcare authorities. In addition, the law provides for the establishment of a National Bioethics Committee for monitoring and consulting purposes. The Committee is composed of 12 members, appointed by regional and national authorities among natural scientists, jurists and bioethicists, and has consultative status.
Finally, genetic testing has impacted on employment issues too. A recent issue of the Spanish newspaper *Expansion* (21 March 2005) revealed that twenty big companies in Spain have offered predictive and susceptibility genetic screening schemes to their top managers. When interviewed, the human resources directors justified the initiative as a way of providing new and crucial information to help their managers with long-term planning in their personal and family lives. 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 request medical consultancy internal or external to the company. According to data provided by the biotech company in charge of this service, so far only one in four managers refused to take the test. The most intriguing issue, however, is that the firms chose to remain anonymous in order to prevent trade unions from seeking to have this service extended to all workers. Whilst the article greatly overestimates the real predictive power of genetic susceptibility testing, it does confirm, at the level of the individual, the tendency to adopt a risk-of-disease approach to health. This reinforces the responsibilisation process which urges the individual to deal actively with this risk and raises the issue of differential access.

**Genetics, healthcare and medical research in Spain: The interviews**

To explore the Spanish context in more depth, a series of semi-structured interviews were conducted across the country with experts in genetic medicine. The ten experts interviewed worked in pharmacogenetics, clinical genetics, genetic epidemiology, health technology assessment and bioethics. The main goal of these interviews was to explore the extent and the implications of the increasing diffusion of genetic testing as it was primarily understood by experts working in different genetic fields. The questionnaire was structured around ten open questions and focused on three main issues: a) the nature of genetic information, b) the current diffusion of genetic testing technologies and c) the contribution of genetics
to medical research and clinical practice. Given the variety of social and political implications at stake, the questionnaire also included a question on public participation, with a specific reference to the role of patient groups. The experts were selected by using the snowball technique, first identifying one expert for each sub-group who was then asked to suggest further colleagues to be interviewed. The sample was obviously far from representative or exhaustive, but all the experts interviewed were leading scholars or professionals in their respective fields and were able to observe the broader phenomenon from a privileged position.

The experts agreed that the advances of medical research in genetics have been remarkable but admitted that so far, the influence on clinical practice has been limited and largely overestimated. Many argued that both the private and the public research agendas have been increasingly focused on genetic aspects of human health, diseases and reproduction, also shifting resources from basic to applied research. This new emphasis has increasingly secured research funds for genetic research, but the geneticists suggested that, compared to the huge potential, this situation is far from satisfactory. I quote:

I am a member of the evaluating committee on research proposals 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. (Clinical geneticist 1)

In contrast, the genetic epidemiologists complained that the situation has 1) sensibly reduced public funding of epidemiological studies on complex diseases, in which the genetic profile of individuals plays a limited role and 2) provoked an excessive emphasis on the genetic components of complex diseases, inducing medical research to neglect a variety of social and environmental factors. They in fact stressed the huge impact of life habits (like eating, smoking and drinking) and polluting agents present in the environment (such as lead, DDT and dioxin) vis-à-vis the development of common diseases like cardiovascular disorders and diabetes.

Although the experts differed in their emphasis on how much an individual’s genetic profile predisposes them to certain diseases, they seemed...
to agree that it is responsible for different predispositions to the development of a number of common diseases. Yet, the experts share the opinion that genetic reductionism, which holds a person’s genes responsible for the development of the major common diseases, is both scientifically flawed and ideologically characterised. First, it is scientifically flawed because recent developments in genetic research show that the genome does not work in a deterministic way. Rather it is largely flexible and is part of a complex network 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. (Clinical geneticist 2)

The genetics-based perspective is also ideologically characterised as it overemphasises the role of individual genetic predispositions which overshadow the social and the environmental factors, thereby avoiding 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. Genetic variation, at least at the population level can neither explain such health differences nor the historical trends. (Genetic epidemiologist 2)

There is a constant exaggeration of the genetic causes of 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 polit-
ical 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. (Genetic epidemiologist 1)

However, a number of experts agreed that medical genetics may yield  
excellent clinical results by focusing on the actual interaction between  
the individual genome, life-time genetic alterations and environmental  
facets, 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, for instance, we have the project INMA  
(infancia e medioambiente) which studies, amongst other things, the  
interaction between environmental pollutants and genetic variation in  
children from before birth to teenage years. (Expert in health techn-
ology assessment)

Modern biology is gradually reaffirming the crucial role of environ-
mental factors in the regulation of genetic expression. In fact, to better  
understand the interaction between the genome and the environment  
we need to integrate epigenetic and classical genetic mechanisms.  
(Genetic epidemiologist 2)

It is really important, in the next ten years, [to work out] 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. (Clinical geneticist 3)

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, except where we are talking about specific monogenic diseases, for which the genetic information is much more relevant. (Expert in pharmacogenetics)

However, the experts suggested 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 suggested that access to genetic information should always be granted to scientists and researchers in order to ensure further medical advances, but that public access should be highly restricted, especially for entrepreneurs or insurance companies, because it can be erroneously used as a discriminating risk factor (Expert in preventive medicine and public health).

The topic of access to genetic information also brought to the fore the issue of patenting and benefits in relation to the socialisation of research costs. One expert in particular expressed concerns about using private genetic information, initially obtained for medical reasons, to develop tests, treatments or products later protected by a patent. In this expert’s opinion, the 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 initial genetic information.

It is not acceptable that genetic information obtained at no cost from affected families turns into a source of commercial profit that imposes enormous costs on the people affected by the same disease. When the genetic information is turned into successful scientific research and an important gene is discovered, these results should be freely accessible for everyone. Unfortunately, it doesn’t work like this: the results of research lead to the development of a specific diagnostic test for the discovered gene, which then gives rise to commercial exploitation. (Clinical geneticist 3)

Some studies reviewed earlier suggested that the diffusion of genetic medicine may be inducing European healthcare systems to switch from preventive to predictive medicine. Discussing these issues in the interviews, some experts agreed that medical research efforts are increasingly
focusing on predictive medicine, but none of the experts interviewed detected significant changes in ordinary medical practice. Yet one expert pointed out that in some cases, for example with breast cancer, the uptake of some genetic testing into the ordinary healthcare system did not produce any beneficial outcomes, neither at the research nor at the therapeutic level.

I believe that a 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 be 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 BRCA 1 to delay or avoid the development of the disease and we found very little work done, [and it was] generally of a very poor quality. (Clinical geneticist 2)

With regards to the interaction between genetic predispositions and unhealthy life-styles, the experts were asked to comment on the possibility of sanctions for the citizens at risk for both genetic and lifestyle factors. Without exception, they approved of more active participation by patients in their health management, but clearly rejected the idea of sanctions.

In contrast, there emerged significant divergence among experts in relation to public participation in decision-making about scientific research and medical policy. Scepticism about public participation, however, was not due to the alleged lack of public technoscientific expertise but rather to an open acknowledgement of the ultimately political nature of the issues at stake. Often the restricted access to the regulatory process was not justified on cognitive grounds but on political ones related to the conflict of interests of patient groups and to their lack of representativeness. Similar observations, though, were generally not raised against business companies, which, given their propulsive role, were perceived as ‘part of the game’.

In contrast, the majority of the experts suggested that patient groups should merely mediate between the scientific community, medical personnel and the general public. Their practical expertise may be crucial in promoting public awareness of the problem, raising funds, lobbying for recognition
of the medical needs of their members and finally in drawing the attention of researchers and healthcare administrators to specific health issues. However, one geneticist suggested that patient groups may also make a crucial contribution during the first stages of medical research, because of their practical expertise in the daily management of given diseases.

There were, once again, interesting differences between clinical geneticists and genetic epidemiologists. Although they encouraged public participation at the consultative level, the experts closer to clinical genetics tended to adopt a deficit model approach to justify the reduction of the number of actors participating in the regulatory 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 the technical and budget issues well. Otherwise there is a risk that proposals approved by parliament on the initiatives of lawyers and bioethicists, who know nothing of the reality of a hospital or laboratory, will not find financial support later. (Clinical geneticist 1)

While acknowledging that the ultimate decision on biomedical research should always rest with elected bodies, the 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 for the political decision-makers, in the sense that if the committee opinion is favourable to the development of a research project, the political authority may still accept or reject it, but if the opinion is negative the research project cannot be later approved by the political authority. (Bioethicist)

In contrast, epidemiologists approved of larger public participation at all levels, including the regulatory one, arguing that an overemphasis on
genetic aspects of health excessively individualises medicine, neglecting the social and environmental factors which affect us all and which should thus be considered and addressed by as many actors as possible.

Conclusion

Drawing inspiration from some pioneer studies on biomedicalisation and geneticisation, this chapter has conducted an exploratory analysis of some of the social and political dynamics associated with the diffusion of genetic testing technologies in medical research and healthcare practices. Drawing from a diverse set of legal and empirical sources and from semi-structured interviews with Spanish experts, this exploratory study concludes that, in Spain, geneticisation processes are currently affecting the medical research agenda more than they affect medical practice, with the remarkable exception of reproductive medicine.

Although the potential fears surrounding individual geneticisation and discrimination resulting from disease susceptibility genetic testing have not yet materialised, the growing influence of genetic studies in the public research agenda is giving rise to a new form of geneticisation. This reduces the efforts made in non-genetic types of medical research, concentrates research on genetic factors and Mendelian mechanisms and neglects the impact of pollution, toxic agents, and of environmental and social factors. Although it may be peculiar to the Spanish context, this result runs counter to many approaches that look for evidence of genetic reductionism and discrimination at the downstream level. In contrast, the data presented suggest that the emergence of an undetected form of genetic reductionism is, in fact, occurring at the upstream level of research orientation.

In reproductive medicine, however, medical practice has been significantly affected by the introduction of pre-implantation and pre-natal screening. On the one hand, reproductive medical practice increasingly seems to endorse ‘weak eugenics’ outcomes. On the other hand, there has been a shift in public health schemes from primary to secondary prevention, especially with regard to those pathologies that cannot actually be prevented, like monogenetic hereditary disorders. In other fields of medical practice, future changes may be triggered by the recently approved Law on Biomedicine.
(2007), which set up a very liberal regime that may facilitate a gradual introduction of new genetic tests in both research and clinical practices.

These outcomes clearly suggest that geneticisation is a complex phenomenon, which cannot be understood simply in terms of a stronger medical emphasis on human genetic characteristics at the expense of the social ones. Geneticisation, rather, emerges as a key dimension of an evolving policy context, in which the social and medical realities are constantly negotiated by the interaction between prospective developments of genetic technologies, emerging research priorities, economic interests, healthcare budget constraints, therapeutic perspectives and medical practices.

My interviews further confirm this complex picture. First, the experts agree that genetic information delivered by susceptibility genetic testing is not clinically but socially exceptional. As the experts suggest, the peculiarity of these genetic data does not depend on their real predictive power, but on their permanent and shared nature and on the social perception and use by third parties. Second, the interviews show an interesting divergence between clinical geneticists and genetic epidemiologists. For instance, whilst the bioethicist and the clinical geneticists frame the diffusion of genetic testing merely in terms of individual consensus, respect for individual autonomy and privacy, the genetic epidemiologists stress the social and even political implications of some genetic testing technologies. These findings suggest that genetics and bioethics may mutually reinforce each other on the basis of a shared individualistic approach to medical and ethical/legal problems (Petersen 2006). Moreover, as the Law on Biomedical Research indicates, the diffusion of genetic testing technologies endorses the neoliberal patient-choice healthcare model, which, in turn, favours a consumerist approach to medicine and healthcare (Henderson & Petersen 2002; McAfee 2003). These results also confirm the mutually constitutive relationship between genetics and the neoliberal narrative (Goven 2006; Rouvroy 2008).

Although generally reluctant to extend decisional power to civil society actors, genetic epidemiologists are aware of the ultimately political nature of the healthcare governance system and are more supportive of general public participation. At the stage of the agenda-setting process, genetic epidemiologists are more supportive of the inclusion of civil society organ-
isations whilst clinical geneticists are more inclined to restrict access to industry representatives and, with due limitations, to patient groups.

The emerging confrontation between genetic epidemiologists and clinical geneticists also highlights the importance of social and environmental factors in the development and treatment of complex diseases. When the epidemiologists suggest that current approaches in medical research overemphasise genetic factors at the expense of environmental and social ones, they explicitly mention the political nature of this shift. As a result, they not only insist on a more comprehensive approach to medical and clinical research, they are also calling for wider participation of civil society organisations and the lay public in general. Pointing to the political aspects involved in current changes in research priorities and healthcare policy, the epidemiologists pave the way to a new understanding of the process of geneticisation, no longer merely confined to medical practice but now widely affecting the society at large.

If the sociopolitical context in which technologies are implemented has a significant bearing on their operation (Goven 2008), then these results reveal that medical expertise in genetics, far from being a homogeneous field, is a locus of conflicting ethical and social perspectives, which engage in power struggles and intersect current political and social phenomena in various ways. They also encourage future studies on the social and political dynamics associated with the diffusion of GTTs to explore their impact not only downstream, in relation to the transformation of healthcare policies, but also upstream, in relation to changing research agendas.

Note

1 Based on the belief that certain disabilities involve unacceptable suffering, the promotion of technologies for reproductive selection is justified on the basis of reproductive autonomy and is not meant to have eugenic goals. However, Shakespeare emphasises that a combination of individual decisions to terminate pregnancy – in a context where social and professional pressures strongly favour this outcome – may result in a situation where, in practice, population-level improvement is achieved via consistent, non-coercive individual choice. The enhancement of reproductive choice and autonomy remains the legitimate goal, but often eugenic trends have been the unintended outcomes.
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