The Birth of a Genetics Policy

Social Issues of Newborn Screening

JOËLLE VAILLY

THE BIRTH OF A GENETICS POLICY

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Social Issues of Newborn Screening

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List of Abbreviations

AFDPHE	*Association française pour le dépistage et la prévention des handicaps de l'enfant* [French Association for Screening for and Preventing of Handicaps in Children]
AFLM	*Association française de lutte contre la mucoviscidose* [French Association against Cystic Fibrosis]
AFM	*Association française contre les myopathies* [French Association against Muscular Dystrophy]
CAF	*Caisse d'allocations familiales* [National Family Allowance Fund]
CCNE	*Comité consultatif national d'éthique pour les sciences de la vie et de la santé* [The National Consultative Ethics Committee for Health and Life Sciences]
CF	Cystic Fibrosis
CFTR	Cystic Fibrosis Transmembrane Conductance Regulator
Cnamts	*Caisse nationale d'assurance maladie des travailleurs salariés* [French National Health Insurance Fund for Employees]
CRCM	*Centre de ressource et de compétence de la mucoviscidose* [Centre of Resources and Competence in Cystic Fibrosis]
DGS	*Direction générale de la santé* [Department of Health]
DHOS	*Direction de l'hospitalisation et de l'organisation des soins* [Department of Hospitalization and Health Care Organization]
DRASS	*Direction régionale des affaires sanitaires et sociales* [Regional Department for Health and Social Affairs]
EBM	Evidence-Based Medicine
ERCF	European Epidemiologic Registry for Cystic Fibrosis
NSCF	Newborn Screening for Cystic Fibrosis
ONM	*Observatoire national de la mucoviscidose* [National Cystic Fibrosis Observatory]
RCT	Randomized Controlled Trial
VLM	*Vaincre la mucoviscidose* [Beating Cystic Fibrosis]
WHO	World Health Organization

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Preface

Fifty years ago this year Robert Guthrie, an American doctor, invented a technique for collecting blood samples from newborns on filter paper in order to carry out tests, thus allowing thousands of children to be protected from the harmful side effects of phenylketonuria, a rare genetic disease. In 2013, more than 50 conditions are now screened for at birth in the United States, most of which are genetic. An important stage in this rapid development was the 2005 report by the American College of Medical Genetics recommending screening for a panel of 29 core conditions and 25 secondary conditions. Today, certain States in the United States screen for diseases that affect people during their adult life as well as for little known conditions or those in which early screening presents limited benefits. Moreover, recent developments in rapid DNA sequencing, and the so-called 'chip' method, mean that genome sequencing of newborns is not such a distant possibility. In a context where screening meets with substantial public approval, particularly from parents of sick children, several national debates are now beginning to consider these genome-related technologies in terms of adapting them to newborn screening. At the same time, observers raise questions about the kind of information that should then be provided to parents, given the large quantity and considerable complexity of the data collected. As we shall see in this book, questions that arose due to changes in newborn screening find a new source in these genome-related techniques, and may even increase as a result. This book focuses specifically on newborn screening for cystic fibrosis (NSCF)[1] in France and many of the arguments that were successful in the debate about this screening programme are now called upon by those advocating an increase in newborn screening more widely. This screening programme for cystic fibrosis (CF) called directly upon DNA analysis in the case of an initial positive test, showing that genetic testing has already entered into the routine of clinical practices and public health.

Therefore, rather than focusing on spectacular medical practices such as new techniques of assisted reproductive technology or so-called synthetic biology, this book looks at seemingly ordinary practices linked to the medical genetics of today; practices that are advancing and yet remain outside of the limelight. However, despite the fact that newborn screening has increased considerably in both the United States and Europe, relatively few social science studies have looked at this question so far. The present study approaches this topic in an entirely new

1 When abbreviations involve French institutions or organizations, the French title is given along with the English translation.

fashion. It analyzes all at once the scientific knowledge, political dimensions and moral questions related to newborn screening, while also examining the interplay between these elements. The methodological framework for the study also takes an original form. It brings together ethnographic observation and sociological analysis, as well as questions from broader spheres. In doing so, this book creates a dialogue between biomedical anthropology, science and technology studies, sociology, and political and moral anthropology, while also fostering exchanges between the Anglo-Saxon and French social sciences.

Of course, certain aspects vary according to social and political contexts, and understanding how these practices are instituted in France may shed some light on certain differences between countries, such as how care provision is organized and the legal framework in place. That being said, there are striking similarities between countries in the field of biomedicine, where knowledge and arguments circulate quickly. Three examples among those outlined in this book can testify to this. First, a study carried out in the United States had a significant impact on the French decision to screen for CF. Second, on a European level, a consensus conference took the same position as the French paediatricians regarding so-called borderline forms of cystic fibrosis. Finally, the decrease in incidence of cystic fibrosis related to newborn screening that has been observed in France has also been noted in certain regions of the United Kingdom and Australia. Above and beyond newborn screening itself, this book looks to offer an account of what screening can tell us about broader societal changes. Indeed, the key issues analyzed in the context of France – evidence, government, norms, the links between the social and the biological, etc. – are also of much wider relevance.

Finally, this book is likely to be of interest to those who find that Michel Foucault's work provides fertile ground upon which to develop their own analyses. His work is used here as something of a 'tool box', as he himself recommended, while taking care to remain rooted within the social dynamics of the early twenty-first century – in other words, taking care to avoid fixing any form of Foucauldian dogma (which would be a somewhat inappropriate homage to a philosopher who displayed such concern for dynamics). Moreover, as we will see, this flexible approach makes it possible to develop a critical perspective that remains respectful of the actors involved, who often find themselves perplexed in the face of their own practices.

I would like to end these preliminary remarks with a few words of thanks. I would particularly like to express my deepest gratitude to Didier Fassin for his consistently sound advice and his constant attention, which both testify to his great academic and personal qualities. My thanks also go (in alphabetic order) to Jean-Paul Gaudillière, Thomas Lemke, Vololona Rabeharisoa and Didier Sicard for having agreed to read and comment on the study that led to this book, despite their extremely busy timetables. I am also very grateful to Vincent Boissonnat, Hélène Bretin, Cécile Ensellem, Boris Hauray and Carine Vassy for our many discussions. Thanks also to Anne-Claire Baratault for her invaluable help with documentary resources and to Lucy Garnier for her care and attention in translating this book. I

would also like to thank the health professionals who gave generously of their time to answer my questions and enable my observations with a view to enlightening my research. I am also indebted to the different people, whether family members with sick children or not, who kindly agreed to talk to me about their lives and allowed me to be present during medical consultations. I would also like to express my gratitude to the members of the jury for the 'Le Monde' Prize for university research, who gave visibility to my work and to Marion Colas from the Presses Universitaires de France (PUF) who assisted me in the process of publishing this English version of my book *Naissance d'une politique de la génétique. Dépistage, biomédicine, enjeux sociaux*, published in French in 2011. Finally, I would like to dedicate this work to the people close to me who have given me strength, and who continue to do so, particularly the strength to change direction when necessary.

Joëlle Vailly, Paris, 20 February 2013.

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Introduction

> We are in a modern maternity hospital; uniformed staff move busily about. We can hear a voice saying, 'Now, only seconds old, the exact time and cause of my death was already known'. A drop of blood is taken from a newborn. A nurse in green hospital scrubs reads printed sheets emerging from a computer: 'Neurological condition, 60% probability; manic-depression, 42% probability; attention deficit disorder, 89% probability; heart disorder'... She looks over at the father... '99% probability. Early fatal potential. Life expectancy: 30.2 years'. 'Thirty years', the father murmurs.//
>
> We see a little boy running in a garden. His mother calls to him; he falls. A voice says: 'From an early age, I came to think of myself as others thought of me: chronically ill. Every skinned knee and runny nose was treated as if it were life-threatening'.//
>
> The parents and the little boy are in a very clean, extremely modern hospital. From the back we see a man behind a desk, explaining: 'Your extracted eggs, Marie, have been fertilized with Antonio's sperm. After screening, we are left, as you see, with two healthy boys and two very healthy girls. Naturally, no critical predispositions to any of the major inheritable diseases. All that remains is to select the most compatible candidate. First we may as well decide on gender'. The man rises: 'Have you given it any thought?' He sits down facing the parents. The mother: 'We would want Vincent to have a brother, you know, to play with'. The man smiles at the little boy sitting on the floor playing with the molecule models. … The man: 'You have specified: hazel eyes, dark hair and fair skin. I have taken the liberty of eradicating any potentially prejudicial conditions: premature baldness, myopia, alcoholism and addictive susceptibility'. The father moves his head; the mother looks at him. The man continues: 'Propensity for violence, obesity, etc.'. The woman intervenes: 'We didn't want... I mean diseases, yes, but...' The father: 'We are just wondering if it's good to just leave a few things to chance'. The man: 'You want to give your child the best possible start. Believe me; we have enough imperfection built in already. Your child doesn't need any additional burdens'.

This is how the future of genetic screening was imagined in the North American film *Gattaca*, which came out in the late 1990s to considerable box-office success. The near-future depicted in the film was characterized by several notions: screening for a series of traits at birth, personalized life expectancy estimates, embryo choice and prenatal selection of genetic traits. This crude entertainment-industry vision of

the issue sought to highlight the dangers of scientistic totalitarianism in a fantasy vision of the future. Let us see how recent history and developments in medical genetics gave a foothold to the film's projections.

In the second half of the twentieth century, biologists considered the genome – that is, the sum of an individual's genetic material – to be a programme that controlled the cellular events and characteristics of living beings. Although some geneticists were cautious in their statements, genetics was nonetheless supposed to influence how we conceived of a wide range of questions: abilities, handicaps, social problems, family relationships and the quality of life (Conrad and Gabe 1999). When the human genome sequence was established at the turn of the millennium, geneticists, particularly in North America, used rhetoric infused with a sense of the sacred, referring to the 'Holy Grail' and the 'Bible', thereby bringing social science researchers to speak of the 'DNA mystique' (Nelkin and Lindee 1994). However, it was precisely this sequencing that shook their certainties, for it did not provide the keys to the secrets of life that most of them had naively hoped for. The very concept of 'gene' became fragmented (Fox Keller 2000). It became necessary to develop more complex and less deterministic models based on the general notion that genetic and non-genetic factors mutually affect one another (Atkinson, Glasner and Lock 2009, Lock and Nguyen 2010). This shift towards greater complexity has given rise today to projects involving large banks of samples in order to understand the respective and combined roles of DNA and the environment in the etiology of multifactorial diseases. Research on 'epigenetics' is now developing, focusing on the effects of the environment on how DNA is used by cells and the inheritability of these changes in the absence of modified genetic sequences (Niewöhner 2011). Moreover, the range of psychiatric and psychological conditions considered to have a genetic component is widening. The molecularization of the living being is thus pursuing its path, integrating greater levels of complexity along the way, and the attention paid to DNA continues.

More broadly, the constituted knowledge called genetics spreads – albeit only in part – throughout societies. Even without taking into account new practices such as paternity tests, choosing embryo sex on the Internet, police identification methods and genetic information on people's origins, which are beyond the scope of this book, the dissemination of genetics knowledge manifests itself in several ways. Patient organizations and the media have their own ways of propagating knowledge about genetics, and the related hopes. In France, the *Association Française contre les Myopathies* (AFM – French Association against Myopathies), among others, actively contributes to 'concern about genes' in the population at large (Rabeharisoa and Callon 1999), thanks to the famous Téléthon, a television fund-raising event calling on viewers to show solidarity and compassion to patients with rare genetic diseases. The Téléthon mobilizes tens of thousands of volunteer workers and collects nearly 100 million euros every year. Anthropologist Paul Rabinow (1999: 39) has described the period during which, for the president of this patient organization, 'the "gene" became the key symbol, the embodiment of fate, the evil locus from which arose death and ruination of innocent life,

and, simultaneously, the site of hope.' Obviously, the co-production of genetic culture within societies also affects decision-makers and political leaders. In this regard, it is of interest to recall the controversy triggered among biologists and sociologists alike by remarks made by presidential candidate Nicolas Sarkozy during his 2007 election campaign.[1] Sarkozy's words, which are consistent with a current of thought that is usually better represented in the United States than in France, expressed belief in the idea of a genetic predisposition to paedophilia and suicide. In sum, people are born paedophiles or suicidal, a representation that de-socializes those behaviours or acts. Private companies have joined in to defend and take advantage of that general understanding. In a context of internationally circulating ideas, knowledge, methods and biological materials, company investments in biotechnology have made great leaps in wealthy countries and the so-called developing countries alike (Rose 2008). In 2011, forty or so companies were selling genetic tests on the Internet (Ducournau and Beaudevin 2011). For a few hundred dollars (the average fee), they analyze their customers' genomes and inform them – so they claim – about predispositions to a great variety of diseases and traits: cystic fibrosis, Tay-Sachs disease, but also breast cancer, susceptibility to heart attack, obesity, in some cases manic-depressive tendencies, hyperactivity, alcohol-dependence, having an above-average IQ, etc. Meanwhile the press takes up stories of customers who have been reassured on certain points – 'I'm not at higher-than-average risk for cancer' – and made to worry about others: 'I'm likely to develop a degenerative eye disease.'[2] There is also a market for pre-pregnancy genetic tests: for $350 a company in the United States called Counsyl will screen would-be parents for a set of 100 diseases but in doing so neglect the question of the diversity of possible conditions. These various tests, openly for sale on the Internet, elicit opposition from some geneticists, who contest their scientific validity and express concern about their psychological and social effects. As early as 2007, the Council of Europe, France's *Agence de la Biomédecine* and the French health ministry, likewise alarmed, organized a conference to analyze the implications of this development and determine what to do about it. Such tests are not permitted in France as they do not comply with French 'bioethics' laws, which, as we shall see, strictly regulate the principles on which genetic testing may be done and genetic tests used.[3] Of course the fact that they can be purchased today

1 Candidate Sarkozy's remarks were as follows: 'For my part, I would be inclined to think that one is born a paedophile – and the fact that we don't know how to treat that pathology is a problem. There are 1,200 to 1,300 young people who commit suicide every year in France, and not because their parents didn't take good care of them! But because, genetically, they had a vulnerability, a pre-existing distress' (see 'Dialogue entre Nicolas Sarkozy et Michel Onfray' in *Philosophie Magazine*, March 2007).

2 Cf. 'Ton génome pour 1 000 dollars,' *Le Monde*, June 7, 2008, and 'Hints of future health from a drop of saliva', *The New York Times*, Dec. 8, 2007.

3 Various French national authorities for regulating biomedical practice have recently put forward proposals for how to frame these practices legislatively.

on the Internet does not say anything about their future. We cannot know what will happen when their limitations as predictors and their potential negative effects on individuals are better known or, conversely, when they start getting greater publicity and the price goes down (in the case of a company called 23andMe, the cost fell quickly from $1000 to $400). Still, this example of the way genetics is developing illustrates that there are strong social dynamics at work. In sum, biologists, those who fund them, patient advocacy groups, biotechnology firm economics, the media, political officials and many other actors are fully implicated in a way of thinking where the inclination is to find explanatory causes for diseases and to orient social questions in the direction of genetics, yet the situation appears relatively complex due to fears of scientistic manipulation on the one hand and hope for cures on the other (Vailly 2013). This duality should be understood in relation to the fact that these new practices are situated at the intersection of science, health and life (Vailly, Niewhöner and Kehr 2011).

Precisely because these developments give rise to fantasies, it is useful to analyze not only what science and medicine *might be able to do* but what they are actually doing in this area today. In this spirit, my book makes use of anthropological and sociological approaches and is therefore rooted in practice. Taking up an idea defended by Michel Foucault, I have sought to view the present up close without falling into a 'theatrical declaration that this moment in which we exist is one of total perdition, in the abyss of darkness, or a triumphant daybreak', but understanding it instead to be 'a time like any other, or rather, a time which is never quite like any other' (Dean 1999: 43). Genetics has a certain propensity to elicit studies that are either extremely alarmist or excessively enthusiastic about current or contemplated practices, or that focus on questions that have already sparked widespread public debate. The investigation on which my study is based is distinguished both by its approach and its type of research focus. It concerns a rapidly developing type of practice applied to entire populations: newborn screening. First of all, let me specify that screening by definition involves identifying individuals within a given population who are at risk for a certain disease or disorder (in which case the screening is followed by a conclusive diagnostic test) or who already have the disease or disorder without knowing it (Armstrong and Eborall 2012). Newborn screening has not drawn much attention outside professional circles, despite the fact that it is the most widely used means of detecting genetic conditions. Specifically, I studied newborn screening for cystic fibrosis, and we shall see why this is an emblematic example of the way norms and techniques in this area are changing. In other words, while seeking to steer clear of false assumptions, my book aims to account for ordinary practices and discourses in medical genetics at the start of the twenty-first century – although the questions it asks are much less ordinary – and to show what screening can tell us about the ways that medicine and our societies are currently changing. But before moving to the heart of the matter, it is important to present the set of analytical tools that will enable us to identify clearly what is at stake.

The Importance of Health Issues and How Those Issues Are Evolving

Questions related to the body, health and well-being are a central preoccupation of contemporary Western societies. This is illustrated by recent fears of epidemics – such as H1N1 flu – as well as increases in screening practices, and the numbers of treatment and well-being centres. This general trend is related to a still more general process of medicalization, by which all sorts of questions become medical ones, including those that used to be located outside medicine's area of intervention (Foucault 2001b: 48-49). As anthropologist Didier Fassin has explained (1996), the medicalization process is manifest in the current expansion of the health field in three directions. The first concerns the targets of health policy – that is, its aims and the people or groups of people it is geared towards. Clinical medicine, which used to handle patients' complaints and involve a direct relationship with a doctor, is combined today with preventive medicine focused on risk and the screening of populations, even though certain screening programmes have given rise to debate due to the anxiety that they can create and due to problems of over-diagnosis (Armstrong 2012). More specifically, screening, anticipation and disease prevention practices are simultaneously addressed to populations as a whole and to individuals. Each person within a given population is urged to get screened whenever possible and to behave in ways that will protect them from the risk of disease. Encouraging individuals to take care of themselves and to exercise vigilance over their own behaviour can work to transfer responsibilities in highly significant ways. Problematizing the issue of lung cancer by emphasizing an approach in terms of genetic predisposition will not target the same people as emphasizing the responsibility of the smoker or occupational exposure to carcinogenic substances. In the first case, the 'target' is the family; in the second, the individual; and in the third, employers. And it does not imply the same research campaigns or health policies: the first approach leads to investigating DNA; the second to prohibiting tobacco use; the third to implementing workplace safety measures. In sum, the first point to be made about newborn screening is that it is consistent with, and part of, a general concern about health and the general expansion of screening practices. The second direction in which the medicalization process is moving concerns the people who intervene. This general category of actors is becoming much more diverse and the relationships between the various components more complex. An example can be seen in patient advocacy groups, increasingly likely to intervene in the public arena to make decision-makers and the population at large aware of their problems. With regard to the history of health and patient organizations in France, the case of AIDS has given salience to a model that involves criticizing delegation and what was termed medical 'paternalism' (Pinell 2004a). It is in this context that we must situate users' increasing demand for information on health. Moreover, patients are now increasingly able to obtain that information through their own efforts, particularly through the media and the Internet. The third direction, less directly related to my study, concerns the diversification of the areas in which health policy is applied. This can be illustrated by 'conduct disorder'

in children, today considered a medical problem after long being thought of as mere agitation. Regarding the primary focus of this study, we can say that given the fact that health has become a precious good and individuals are increasingly considered responsible for their own health, it has become morally reprehensible not to take care of oneself and not to prevent a loved one's suffering. Likewise, for anthropologist Margaret Lock (2000), well-being and steering clear of disease have become a sort of virtue – especially, she adds, among the middle classes. For make no mistake, taking care of one's health requires skills and resources that are not equally available to all, and the technicizing of medicine in rich countries is obviously no threat to stratification based on socio-economic inequalities. The observed collective sensitivity to health and life-related issues does not mean we can assume that health care is a priority in all circumstances. What are we to think of a prison population exposed to serious health risks and going untreated, or employees who remained exposed for decades to asbestos in France when the danger had already become clear, or groups of Roma living in unhealthy conditions, and similar problems? This study will partially mirror this last observation, for it analyzes a health policy and programme that concern very few sick people and handle them in a particularly concentrated, active way. It remains to be seen what the conditions of possibility were for implementing this policy. Another apparent contradiction is that although bodies exposed to suffering elicit compassion, this does not mean that people with handicaps have their full place in society. There is indeed a general concern for health but, on the one hand, not all health problems are taken into account the same way, and, on the other, while there is compassion for suffering bodies, the other's physical otherness is not fully accommodated. All of this means that different, and in some instances clashing, elements come into play, which need to be analyzed. Because Michel Foucault's analytical framework enables us to attend at once to knowledge, policies, norms and morality, it will enable us to move forward in this direction. First, however, it is important to specify certain changes that have taken place in the field of health.

The medicalization of people's existences is accompanied today by a movement that sociologists call 'biomedicalization', which affects primarily, but not exclusively, Western countries. This term designates the process by which the techno-scientific approaches associated with the life sciences are being hybridized with those used in clinical practice. This process, which developed during the second half of the twentieth century (Gaudillière 2006), has been greatly accelerating since the 1980s (Clarke et al. 2010). It obviously does not imply that all earlier approaches to medicine have disappeared, but rather that new practices, changes and replacements will gradually be added. One characteristic of biomedicalization, then, is related to the emergence of new knowledge and the effect it has of obscuring the boundaries between sciences, techniques and medicine. Because biomedicine means to be both scientific and medical, it raises issues related to the status and productions of science, and these are of interest to anthropologists of biomedicine as well as sociologists of science and epistemologists. The question of what role should be granted to social processes

versus intrinsic characteristics of nature is debated at length by these thinkers and researchers. Many social science researchers today agree that instead of analyzing science's finished products, the priority is to study science in action, meaning ethnographic studies and studies of science's material forms (Cambrosio, Young and Lock 2000). The point is to investigate the dynamics of science, the social conditions in which it develops, and the way it legitimizes actors and is itself legitimized by those actors' social position. However, precisely because biomedicine intersects with clinical work and implicates people as living beings and their lives, it has its own issues and values that are not necessarily to be found in the sciences *per se*. In other words, because biomedicine applies at once to people, organs, and molecules, it brings into play both 'living beings' (*les vivants*) and life itself (*le vivant*).

An Analytical Framework

Michel Foucault's theories provide keys for analyzing health care questions and the larger issues they raise precisely because they highlight how life and health care have made their way into political strategies. Foucault's notion of biopower refers to the emergence in the seventeenth century of a 'power over life' that was deployed at an individual level and somewhat later at a collective level as well (Foucault 1998). At an individual level, because this power focused on the body for the purpose of training it and increasing its strength by ridding it of bodily, sexual and social habits that might hurt its health; at a collective level in that it aimed to control the species, which then became a stake in political strategies for ensuring proliferation and longevity, strategies grounded in demography and epidemiology. These two elements, which Foucault referred to respectively as 'anatomopolitics' of the human body and 'biopolitics of population', constituted two poles that could also intersect with each other. From the early twentieth century, the desire for health no longer consisted solely in avoiding diseases or premature death but also in seeking to optimize the body so as to attain a sort of general well-being (Rose 2001). The World Health Organization (WHO) went even further, defining health as 'a state of complete physical, mental and social well-being and not merely the absence of disease or infirmity'.[4]

This already enables us to better grasp this process of the biomedicalization of human lives. However, if we were to leave things there, the historical movement I have just briefly retraced would seem somewhat linear. First, it seems oblivious to disparities between the goal of good health and the means available and actually used in societies to reach that goal. In Western countries, this presentation of things would have to be clarified and refined by studying various social worlds;

4 Cf. 'Preamble' to the World Health Organization Constitution adopted by the International Health Conference, New York, June 19-July 22, 1946, http://www.who.int/suggestions/faq/en/.

specifically, worlds marked by social inequalities and unfavourable living conditions, where having enough to eat and a roof over one's head is more urgent than taking care of one's health. Moreover, established as it has been on the basis of various European countries, the process seems to ignore the countries where health is not really taken into account in political or policy strategies. Anthropological studies in South Africa (Fassin 2007) and Brazil (Biehl 2005) are working to expose and illuminate this rather large blind spot. By showing how the political is inscribed in human bodies, Foucault's analytical framework enables us to integrate situations such as social inequalities, divergent interests, etc., that may counterbalance the very real general progression of medicalization. Second, this presentation would have to take into account historical periods in which promotion of health and biological life comes up against other strategies, such as those for waging war. While this question would take us too far afield, this possibility of integration was suggested by the philosopher himself (Foucault 1988). It should also be noted that some authors think that life became a focus of political strategies much earlier than the seventeenth century (Agamben 1998), while others have suggested that the development was triggered by general mechanisms that can be found in other historical periods and places, such as ancient Rome (Fassin 1996). Lastly, it should be stressed that using Michel Foucault's analytical framework implies following the modulations of thought that changed over time.

Indeed, the philosopher later introduced the notion of 'government' and this shifted the focus of his theories of power. He did so at least in part in response to critics who had faulted him for not acknowledging potential resistances to the seemingly omnipotent power he had defined (Gros 1996). With its notions of control, surveillance and the production of docile, submissive bodies, the first version of his biopower theory did indeed leave itself open to this critique. What Foucault added more clearly later was a dimension of freedom. This term immediately calls for two remarks. First, in a crucial way, Foucault's freedom is internal to power, for not only does that power encompass freedom in its own techniques but it can only endure by relying on that same freedom. What bolsters the stability of that power is that it neither represses nor forbids – this explains the metaphor of the bumblebee ruling without using its stinger – but rather urges and produces. Second, the philosopher is not analyzing one or several powers so much as power *relations*, understood to be exercised over a recognized 'other' who is to be maintained as an acting subject. Foucault thus defined government as the set of more or less conscious, calculated modes of action aimed at affecting other individuals' possibilities of acting. In this sense 'government' refers not only to political structures and state management but to a way of structuring others' potential fields of action ('*conduire les conduites*' or 'leading conduct'). Consistent with the fact that power in this conception is not located exclusively 'at the top', the analytical framework includes a 'microphysics' of power, composed of all the apparently minor power processes that get diffused within society. It is therefore important not only to perceive and seek to understand the strategies and instruments of government previously defined but also power at its extremities, where it comes to resemble capillaries. The question of propagating

genetic knowledge in society must therefore be related to Foucault's concept of diffuse, diffused power.

Toward the end of his life, Michel Foucault turned back to his past work, taking as his theoretical focus 'the field of experience', a concept that provided a new foundation for the body of his previous work. Although he did not develop this idea as fully as his theory of government, he did write of it in the following terms: 'In these three areas – madness, delinquency, and sexuality – I emphasized a particular aspect each time: the establishment of a certain objectivity, the development of a politics and a government of the self, and the elaboration of an ethics and a practice in regard to oneself. But each time I also tried to point out the place occupied here by the other two components necessary for constituting a field of experience' (Rabinow 1984: 387). In other words, the field of experience was conceived as a fold situated between a question of truth linked to knowledge, a question of power, and a question of the subject's relationship to the self (Gros 1996). It should be emphasized that because the subject's relationship to the self is conceived as an 'experience' it is not at all metaphysical but necessarily anchored in practices and discourses.

Foucault thus moved from biopower and biopolitics to government, the subject and ethics by reshaping his concepts. My argument here is that these concepts provide a particularly suitable framework within which to study newborn screening and biomedicine. This might be linked to the fact that Michel Foucault was a student of the philosopher and historian of biology and medicine Georges Canguilhem. The fact is that these tools allow several notions to be both reconciled and linked together, which I will of course have occasion to discuss at several points further on. The question of 'forms of knowledge' will be taken up in connection with the role of genetics and statistics in the history and practice of newborn screening for cystic fibrosis; the role of the body and health in policies such as screening will be resituated in the framework of 'biopolitics'; and 'subjectification' – that is, the constitution of choice-making subjects and their dialectical relationship to passive 'objects' – will be analyzed in connection with the notion of 'informed consent', a notion increasingly present in medical law, as we shall see. Equipped with this highly relevant analytical framework, we are now ready to move to the heart of the subject of newborn screening.

Screening Newborns

The screening of newborns, usually carried out to detect genetic conditions, is consistent with the changes described above both in terms of politics and policies of life and the living being, medicalization and disease prevention, and in terms of the development of biomedicine and the genetic approach. It also involves the question of how public policy handles issues involving young children. During the twentieth century, children were the target par excellence of screening-based medicine, through vaccination and health education campaigns, as well as the study of children's physical and mental development (Armstrong 1995). The practice of

keeping track of how newborns are developing reflects an even more longstanding concern about the health of children in their first months and years of life. Although this concern obviously does not explain everything, it should nonetheless be seen as related to the substantial quantitative development of newborn screening in wealthy countries. As early as 2001, an article published in the prestigious biomedical journal *Science* made the following announcement: 'The ability to scan one sample for some two dozen inherited disorders is about to cause an explosion in neonatal screening; few health systems are prepared for the consequences' (Marshall 2001: 2272). In the United States, more than four million newborns are screened every year. Today no fewer than 50 conditions are screened for on average in the country's various states – rare diseases, as most affect between one child in several thousand and one child in several hundred thousand.[5] In Europe as I write, Germany screens for 14 diseases at birth, while Great Britain screens for five and France for six.[6] This has brought about two developments. First, the standardization and routine use of new techniques mentioned in connection with the rise of biomedicine has made it possible to use those techniques on large populations and to analyze a greater variety of conditions. Spectrometry, chromatography and direct study of certain sections of the DNA chain have led to the practice of analyzing a great number of proteins and genetic anomalies using a few drops of a newborn baby's blood. Second, these new screening practices are modifying some of the legitimacy criteria for newborn screening that had hitherto been in effect. Until about 15 years ago, newborn screening had to meet a set of criteria known as Wilson-Jungner (Wilson and Jungner 1968), established by the WHO in 1968 and reapproved by a North American consensus conference in 1988. Among other things, these criteria defined disease or disorder characteristics (the disease has to be known, serious, etc.), the way diseases might be identified (signs and markers have to be reliable, etc.), public health care requirements (screening cost should not be too high nor exceed the cost of treating the sick identified, etc.) and the information provided to screened populations (they have to be clearly informed about the test and cannot be tested without their consent). Among the criteria relating to the characteristics of the disease, the existence of a cure was considered to be a prerequisite. Gradually, however, this has been replaced by the idea that there must be benefits to treating the disease or the condition early, even in cases where, at the existing state of medical knowledge, there can be no return to normal health. Obviously the decisive point here is what is meant by 'benefits' and 'treatment'. Moreover, the fact that it was in parents' interest to be informed of any disease

5 For further information on newborn screening in the United States, see the site of the National Newborn Screening and Genetics Resource Center, http://genes-r-us.uthscsa.edu/.

6 Newborns in France are screened for the following diseases: phenylketonuria, hypothyroidism, congenital adrenal hyperplasia and cystic fibrosis (CF). In France's overseas *départements* and territories, screening for sickle cell disease has been generalized while in mainland France it is restricted to populations of African origin or from the Mediterranean basin. Screening for deafness was added to this list in 2012.

their newborn might have began to be taken into account, independently of the benefits the children themselves might draw. Despite these changes, until recently there has been little social science study of newborn screening compared to the attention given to genetic diagnoses of the foetus and what are known as 'genetic counselling' practices aimed at informing people about the origins of a disease, the risk of transmission, and the possibilities of detecting a potential genetic disorder in the foetus.

My research examined newborn screening for cystic fibrosis (NSCF). This genetic disease, which can vary greatly in severity, is primarily characterized by usually serious respiratory disorders, due to the accumulation of a viscous mucus that causes infections, as well as digestive disorders. A salient feature of the disease is that it is both rare and the most frequently occurring monogenic disease in populations of European origin.[7] It is estimated that in France one out of 4,400 people is born with CF; in France today there are approximately 6,000 people with the disease. Some of these aspects – severity, relative frequency – amount to factors that work to mobilize social actors around the disease. Above all, CF fits into the fundamental socio-historical developments by which chronic diseases became a priority concern in Western countries (whereas up until the Second World War, these countries were primarily affected by acute diseases). Biologists and geneticists have devoted and continue to devote much research to CF at the international and national levels; there has been much hope among both researchers and patients and their families that a form of gene therapy will be found for this disease; that is, a therapy that will correct the defective gene in the sick person. It is in this context that in 2002 France launched nation-wide NSCF. Two points should be made in this regard. First, France occupies an important position in that it was the first country to have adopted this practice, along with much of Australia and New Zealand. Second, NSCF is emblematic of newborn screening and how it is evolving, first of all due to the fact that there is no known cure for the disease. While patients' life expectancy has risen by 15 years in the last approximately 15 years, it is no higher than 40 years in Europe. The absence of curative treatment was (and to some extent continues to be) an issue in the debate in biomedical and health administration circles on whether NSCF was a good idea and whether and how it could be justified, given that the benefits for children who screen positive had not been clearly established (MMWR Recommendations and Reports 1997, 2004). An exploration of biomedical literature shows that this debate has been medical – Does NSCF offer enough patient benefits given its drawbacks? – and/or scientific – Is there scientific proof of those benefits? – and/or moral – Is it ethical to take into account possible benefits to parents? I will of course be returning to these questions. Today the cursor seems to be shifting in favour of NSCF, and other countries, including the United States and Britain, have begun the practice. There is also a

7 A monogenic disease involves one major gene, although other genetic factors as well as social and environmental ones can affect how serious the disease is.

techno-scientific indication of NSCF's position at the transition point between already established and new newborn screening tests. The technical aspects of the practice will be specified below, but here it is important to know that NSCF involves direct study of the DNA of people who have tested positive once – and this is the first time that such study has occurred on this scale in France. Direct DNA study may induce various effects that would not occur with other biomedical practices. It may work to diffuse particular representations related to the image of DNA (Nelkin and Lindee 1994); it may focus family ties on biology rather than emotional relations. And in France it is subject to stricter legal regulations than traditional biological tests: it requires written consent of the concerned party – in this case, the newborn baby's parents.

At a time when genetics is undergoing the important developments indicated above, it is worth analyzing the social issues raised in connection with an actual policy and real practices. The point is to study what that policy and those practices can tell us about the fact that the issue of the living being and life has become part of political strategies and about wider societal changes. What are the defining characteristics of a policy of the living being (*le vivant*) or living beings (*les vivants*), that uses the form and content of the knowledge called genetics? Under what scientific and social conditions is such a policy possible? What does it tell us about the politics and government of people-as-living-beings? How is it involved in producing norms and values? In the present study, the point is to analyze *as a field of experience* (in the sense defined above) a *policy that developed out of medical genetics*; that is, to analyze it three-dimensionally: *in scientific*, *political and moral terms.* The aim, then, is less to assess the benefits of the policy and point out its limitations than to analyze the conditions under which it is possible to implement such a policy, the varieties of political logic underpinning it, and its effects on norms and values. In doing so, I will show in particular that *newborn screening outlines a new political and moral space of genetics*. My study encompasses an apparently heterogeneous set of discourses and practices, decisions, laws, administrative measures, scientific statements and moral propositions, all organized around a field of experience whose three dimensions – scientific, political and moral – may be interrelated. From the outset we can hypothesize that each of these dimensions helps constitute the other two: knowledge affects moral understanding and political practices, etc. This hypothesis will lead us to consider questions that, although certainly not meant as universal, may well reach beyond the frame of biomedicine, questions that may be described as sociological or anthropological knots. Borrowing that term from psychiatry, Ian Hacking (2005) defined a knot as an issue produced by contradictory tendencies. Here those issues are truth (versus seeming), choice (versus being forced), the norm (versus deviance) and a quality life (versus a poor quality life). In other words, we will also be studying a policy of the living being and living beings in the framework of a historically situated social dynamic, determining and defining what it tells us about the more general issues it both reflects and fuels.