Vulnerable populations and genetic disorders: a socio-science approach to the application of genetic technology in China

Sui, S.

Citation for published version (APA):

General rights
It is not permitted to download or to forward/distribute the text or part of it without the consent of the author(s) and/or copyright holder(s), other than for strictly personal, individual use, unless the work is under an open content license (like Creative Commons).

Disclaimer/Complaints regulations
If you believe that digital publication of certain material infringes any of your rights or (privacy) interests, please let the Library know, stating your reasons. In case of a legitimate complaint, the Library will make the material inaccessible and/or remove it from the website. Please Ask the Library: http://uba.uva.nl/en/contact, or a letter to: Library of the University of Amsterdam, Secretariat, Singel 425, 1012 WP Amsterdam, The Netherlands. You will be contacted as soon as possible.

UvA-DARE is a service provided by the library of the University of Amsterdam (http://dare.uva.nl)
CHAPTER ONE

INTRODUCTION

This research is a subproject of the international research project, the Socio-genetic Marginalization in Asia Programme (SMAP), a comparative approach to the relationship between genomics, governance, and social identity. SMAP explores the cultural, social, and economic aspects of the role which genetic technologies have played in the area of state organisation, population policies, healthcare systems, and research regulation in China, India, and Japan. It sheds light on the applications of modern genetic technologies generated in different social contexts (Sleeboom-Faulkner 2005). As a part of this international project, this research takes the application of genetic testing and involved vulnerable populations in China as its point of departure, and explores links with social economics, national population policy, the national healthcare system, and traditional/local culture in China.

RATIONALE OF THE RESEARCH

The development and current achievements of genetics technology enable its application in a growing number of contexts. In fact, the application of genetic testing not only reflects the development of genetics technology but also incorporates socio-political, socio-economic, and cultural characteristics. Ethical codes and notions of professional responsibility in medical activities are perhaps differently understood in various countries because of different
social factors. In order to understand well the current practice of genetic testing and how the practice evolves, one must consider the socio-political, socio-economic, and cultural factors of the societies where they emerge and exist.

In China, good or superior birth has been highly encouraged and the one child policy represents an extraordinary attempt to engineer national wealth, power, and global standing by drastically slowing population growth, with profound effects on virtually every aspect of Chinese life (Greenhalgh 2003). At the same time, China’s socio-economic and healthcare systems are undergoing fundamental change. As China changes from a planned to a market economy, health policy makers are grappling with pressing issues of cost containment, structural reorganisation, and market regulation (Meng 2004). During economic development, social issues related to healthcare are becoming more problematic. There is a nationwide shortage of medical resources and an imbalance of medical resource allocation between urban and rural areas. The high cost of medical care makes it difficult for people with health problems to gain access to satisfactory medical help, especially for low income families and those in comparatively remote and poor areas. Meanwhile, under the circumstances of a market economy, the potential commercial value of applications of genetic technology is coveted by pharmaceutical and biotechnology companies. Commercialised genetic testing is emerging, accompanied by various social and ethical issues that deserve greater attention.

New genetic testing technologies provide diagnostic and predictive genetic information. But this in itself also raises questions about consent, confidentiality, and the use of information by family members and other third parties (Konrad 2005). Psychological harm can also result within a family as a result of genetic information being acquired about individual members. This can create tension and become a source of resentment, hostility, and discrimination. Information about an individual’s genetic status can have much wider adverse implications for a person’s financial security, employment, lifestyle, the welfare of his/her family, and psychological wellbeing (Marteau & Richards 1996). Additionally, the application of advanced genetic technologies is playing an important role in the ‘new eugenics’ of improving the quality of the population within the context of the one child birth policy (Mao 1998; Wang 2004; Nie 2005). It is therefore of great significance to use a social science approach to understand the practice of genetic testing, the issues involved in this field, and its implications in the Chinese context.
OBJECTIVES OF THE RESEARCH

Genetic testing is available nowadays to identify many genetic disorders and is becoming popular in China, especially in the field of reproductive technology such as prenatal genetic testing. It may benefit many different interest groups, such as individuals and families with a history of genetic disorder, pregnant women, employers, and health or life insurance companies. At the same time, as an application of new genetic technology, the growing availability and accessibility of genetic testing in China has had an impact on Chinese society. Some issues have not attracted the attention they deserve, such as genetic privacy, individual autonomy, and reproductive decision-making; and some problems are emerging along with the development of genetic technology and its application, such as genetic discrimination and stigma, and the use of pre-implantation genetic diagnosis or prenatal genetic testing to create ‘perfect’ donor matches for stem cell transplantation. In order to better understand the situation of genetic testing in the Chinese context and the interest groups involved, a study of the social, ethical, political, and legal issues related to genetic testing is required. Based on field research, this study explores these issues. The main objectives of this research are:

First, to map and understand the practical application of genetic technology in the Chinese context;

Second, to analyse the role and actual effect which laws and regulations, ethical guidelines, and national/local health policies play in the practice of genetic testing and its combined service;

Third, the core research aim is to understand the views and practices of various interest groups involved in genetic testing and to delineate the vulnerability of social groups in the Chinese context from socio-economic, political, and cultural perspectives.
MAIN RESEARCH QUESTIONS

This research focuses on the practice of genetic testing in China. From a social science perspective, the research mainly attempts to analyse: how the Chinese social economy, national policies, and culture are embodied in the practice of genetic testing; how social factors impact on the form and application of genetic testing in Chinese contexts; how social factors shape views and actions regarding the reproductive decision-making of involved populations; whether genetic testing sharpens reproductive pressure, especially on females; how the practice of genetic testing reflects systems of medical governance, the distribution of medical resources, the doctor–patient relationship, the vulnerability of populations, and population policies in Chinese socio-economic and socio-political contexts. Additionally, the research shows how medical genetic services play a role in the prevention of birth defects and in the implementation of Chinese eugenic policy.

This research divides genetic testing into commercial genetic testing and clinical genetic testing. Commercial genetic testing is offered by biotechnology companies. The research shows how commercial genetic testing is operated as a business, analyses the advantages and disadvantages involved, and looks at the social and ethical issues that cause – or will be caused by – the commercialisation of genetic testing. Clinical genetic testing is, however, the main emphasis of this research. Through a comparative approach, the differences in the clinical practice of genetic counselling between China and the UK are accounted for. Through the case studies on thalassaemia and Duchenne muscular dystrophy (DMD), the research discusses the vulnerabilities of patients/carriers and their families affected by genetic disorders through the following questions: how clinical genetic testing influences reproductive decision-making and reproductive choices; what difficulties and burdens they encounter in their daily life; and whether social welfare and help are available to them. Additionally, the research raises and analyses the question of how genetic services play a role in the prevention of birth defects and take on the duty to improve population quality under Chinese eugenic policies. Based on these research questions, this study attempts to explore the identity and vulnerability of populations regarding the practice of genetic services in
China, and it delineates the socio-economic and socio-political factors that influence, shape, and sharpen their vulnerability.

DEFINITION AND DELINEATION OF MAIN RESEARCH ISSUES

This research mainly concerns carrier genetic testing and predictive genetic testing. Carrier testing is a method used to identify individuals who carry a genetic abnormality that does not affect the health of the person in question but increases the risk of producing offspring with a serious genetic disorder. Identified carriers can harbour continuing regrets and worries, while others may fail to recall the significance of their test result over time. More constructively, the information may be useful in making reproductive plans (Prainsack 2006; Brandt-Rauf et al. 2006). In practice, in areas of high occurrence for certain genetic disorders, such as thalassaemia in southern China, when considering the prevention of birth defects carrier testing is encouraged in premarital or prostational check-ups. Genetic counselling is recommended before and after the test to prevent confusion over the difference between being an asymptomatic carrier, and therefore not developing any signs of the disease, and being affected by it. Although genetic counselling aims to minimise adverse psychological reactions, it does not prevent carrier screening from creating opportunities for genetic discrimination, as happened with sickle cell screening in the United States in the 1970s (Lemmens 2000; Mc Carrick 1993).

Predictive testing identifies individuals who have inherited a gene for a late onset disease, but who currently appear healthy. A distinction can be made between presymptomatic testing and susceptibility testing. Presymptomatic tests identify healthy individuals who have inherited a defect in a specific gene for a late onset disease, with an almost one hundred percent risk of developing the disease. These tests do not inform about the severity or the time of onset of the disease. Examples of such conditions are Huntington’s disease (HD) and cystic fibrosis, which are monogenetic disorders (single gene disorders). Susceptibility (or predisposition) tests identify individuals who have inherited a genetic variant or variants which may increase their risk of developing a multi-factorial disease (such as bowel cancer, breast cancer, heart disease, diabetes, hypertension, and dementia), due to the interaction of
genes and the environment. Susceptibility tests are mainly offered by commercial biotechnology companies in China.

One problem is that with the current available knowledge, technology can identify those at risk of disease but cannot prevent it or improve their treatment. The benefits of the commercial tests may be limited, given also the risk of the genetic information for people found to be susceptible.

Information about a person’s genetic make-up may be of interest not only to the individual but also to other members of their family, to their employer, their health or life insurance companies, and other possible agencies, including the state (Clarke 1994). Confidentiality and privacy of genetic information is an issue that, if not regulated, could lead to new forms of discrimination (Billings et al. 1992). The identification of those at increased risk for common diseases has the potential to cause distress, denial, and inappropriate feelings of fatalism, and opens up the possibility of discrimination. In fact, discriminatory practices directed against ‘abnormal’ people are greatly feared, and not only ‘abnormal’ persons but also persons with ‘abnormal’ family members may experience problems finding a spouse, amongst other social stigmas. For these and other reasons, the genetic information produced through predictive and carrier tests can potentially harm entire families. Keeping information about familial ‘abnormalities’ secret is thus an important aspect of the maintenance of privacy. Additionally, there is a widely held notion that in Chinese tradition it is not uncommon to withhold the truth about disease from a patient. This raises the question of how this notion is practised in the case of genetic test results.

Prenatal genetic testing generally is done in China during pregnancy either to screen for or diagnose a birth defect. Theoretically speaking, the main goal of prenatal genetic testing is to provide families with information to make informed choices about pregnancy and reproduction, and to assist the woman’s physician in providing the best care and management for her pregnancy (Jorde et al. 1999). Prenatal tests provide valuable genetic information about the health of the foetus. However, the tests involve ethical considerations and social implications, such as informed choice, reproductive decision-making, Chinese population policy, prevention of birth defects, and the cultural values attached to offspring. This research involves case studies of the clinical prenatal diagnosis for a birth defect with
thalassaemia and DMD.

Thalassaemia is widely prevalent in the south of China. Blood transfusion, which is a common treatment for thalassaemia, can control but cannot cure the disorder. A special treatment exists which uses donor umbilical cord blood, however this involves the parents of an affected child having another unaffected child who is genetically matched to provide the life saving cord blood, which can be transplanted into a sick brother or sister. Such a child is called a ‘saviour sibling’. The intentional creation of a ‘saviour sibling’ may lead us to consider further the social and ethical implications involved.

DMD was chosen as a research target for other reasons. DMD is an X-linked genetic disorder, thus it is passed down from the mother, who is a DMD carrier, to her son. This may have an impact on the position of the woman within the family; women are already to a certain extent weak and vulnerable, and this may be enhanced by the traditional Chinese preference for boys that may sharpen the social stigma for and discrimination against families with a son with DMD. DMD is a chronic, serious, and lethal disorder. Children affected by DMD slowly lose the abilities of walking, standing, and turning over because of muscle atrophy. Currently, around the world there is still no medical cure. The life condition for children with DMD and their families may not only reflect the vulnerable situation of those with a genetic disease but also that of the disabled.

Birth defects are structural or functional abnormalities present at birth that cause physical or mental disability. A birth defect is a problem that happens while a baby is developing in the mother’s body. In fact, birth defects are a major cause of infant mortality in many parts of the world (WHO 2006). Birth defects and the disability resulting from them have been considered among the problems that affect population quality and cause heavy financial burdens for families and society (Luo 2005). With the gradual increase in the application of clinical genetic testing, prenatal genetic testing may play an important role in the prevention of birth defects. Meanwhile, some social implications, such as population quality, ideas of ‘good birth’ and ‘bad birth’, reproductive pressure, and the eugenic approach of avoiding ‘inferior birth’ deserve more consideration.
METHODS ADOPTED IN THE RESEARCH

This research was conducted as part of the larger international research project SMAP, which was funded by the Netherlands’ Organisation of Science (NWO). SMAP aimed to identify the socio-cultural domains relevant to socio-genetic marginalisation through empirical research. As a subproject of SMAP, this research adopted empirical fieldwork as its research basis. To develop this research, during a three year period from 2006 to 2009 several empirical field studies were carried out. The details of the study context – fieldwork sites, interviewee selection, and data collection – are presented in Chapter Two.

The methodology of this research was eclectic, and was chosen based on suitability for the research problem under study. A variety of methods was utilised to collect a range of information, used to understand the social context and to analyse research problems.

Archival research was conducted into existing national rules and regulations and international guidelines for the application of genetic technology, the practice of birth control, and the cultural and religious background of the bioethical and social questions related to genetic testing. The written information resources and materials referenced in this research include books, academic papers, newspaper articles and reports, government department documents, codes of conduct and regulations of institutions/organisations, official guidelines and reports, leaflets or propaganda sheets, informed consent forms, written court verdicts of lawsuit cases, and so on. Most of these reference resources were collected from the Internet, libraries, and book stores, and some materials were collected from the fieldwork sites or from interviewees. A literature review of written materials was valuable not only as a learning process to gain theoretical cognition and rational knowledge and understanding of the research domain, but also as a means by which to gather information for the field study to be used to understand the basic situation of genetic tests and the background social context.

In order to get a better insight into the relationship between genomics and society, it is required to understand how genomics affects socio-economic development and religious and
cultural experiences. To gain firsthand information and experience, field research was carried out over three years. Firsthand information was collected by conducting in-depth interviews with genetic counsellors, clinical geneticists, pregnant women, thalassaemia carriers, DMD carriers, the mothers and/or fathers of children affected by thalassaemia, the mothers and/or fathers and family members of children suffering from DMD, the executive coordinator of patient organisations, agents of biotechnology companies, and agents of insurance companies.

Some basic research issues were designed into questionnaires. The questionnaires were semi-structured, and sometimes questions from the questionnaires were also used in the interviews. The questionnaires were used to obtain opinions and attitudes towards genetic testing, the reasons to take or not to take genetic tests, privacy and truth telling, attitudes towards genetic information and pregnancy termination, and so on. These opinions were elicited from various interest groups.

Additionally, the personal experiences of clients were acquired through participant observation in the clinic, for instance by sitting in on clinical genetic sessions, and accompanying patients to observe the procedure of testing and consultations. This also included observing the medical facilities, propaganda posters in the hospital and laboratories, and collecting informed consent forms in the clinics. Participation and observation of target families in case studies helped to understand their daily life context, and the feelings and problems they have in daily life.

**STRUCTURE OF THE DISSERTATION**

In this thesis the objectives, methods and results of the research are presented in eight chapters:

**Chapter One** is the introduction to the research. This chapter presents the rationale and the objective of the research. It defines and delineates the research problems, the fields and issues involved in the research, and it provides a general overview of the research project, the methodology, and the content of the following chapters.
Chapter Two presents the research context. This chapter looks back at the development of medical genetics in China. It briefly introduces the research domain and gives the details of fieldwork sites, interviewee selection, and data collection.

Chapter Three provides an empirical account of the application of genetic counselling in China and makes comparisons with the situation in the United Kingdom. It demonstrates how Chinese practitioners understand and relate to various ethical issues such as non-directiveness and informed choice and decision-making, and explores the extent to which this reflects the administrative, political, and socio-economic environments in Chinese society. It offers insight into the current situation of genetic counselling in China, elucidating the practice of non-professional genetic counselling, the economic conditions which affect genetic counselling, and the lack of full healthcare coverage.

Chapter Four focuses on the situation of predictive genetic testing offered by biotechnology companies in China. As predictive genetic testing is not formally regulated in China, its commercialisation has caused, and will continue to cause, social and ethical problems. Examples include the exaggerated advertisements that mislead customers, discrimination as a consequence of the misuse of genetic information, and the reliability and usefulness of test results and advice. Additionally, the system of business operation and agent recruitment of some biotechnology companies are problematic. This chapter argues that these issues require for more attention to be paid to them by the public, including the involvement of ministerial departments.

Chapter Five delineates the current application of prenatal genetic testing for thalassaemia, and the reproductive decision-making of thalassaemia carriers. It makes observations about the difficulties faced by prospective parents who have a high chance of conceiving a child affected by thalassaemia, and the social dilemmas they encounter. The chapter also analyses some factors affecting the decision to produce a ‘saviour sibling’, including financial implications, the state family planning policy, influential images and information conveyed
through the media and propaganda, the advice and counselling from doctors, psychological pressure from the community, and social discrimination.

**Chapter Six** presents the application of genetic testing and genetic counselling on Duchenne muscular dystrophy and analyses the vulnerability of families with DMD in China. Genetic counselling and testing for DMD has important implications for reproductive and life planning decisions. This chapter offers a better understanding of the economic difficulties, psychological distress, self-contempt, and family stigma that Chinese families with DMD must confront. The chapter also rethinks some related social issues such as genetic discrimination, the distribution of health resources, the impact of genetic technology on reproductive decision-making, and the limited social support and help for vulnerable groups.

**Chapter Seven** analyses the important roles that genetic services play in the prevention of birth defects and in the improvement of population quality in China. It introduces understandings of eugenics, or Chinese *yousheng*, and its relationship to genetic services and the prevention of birth defects. It also discusses issues involved in China, such as public awareness of primary prevention of birth defects, and reproductive responsibility and choice. It points out that due to the national desire to reduce birth defects, issues concerning some individual rights and choices involving personal genetic information and decision-making do not receive the attention they deserve.

**Chapter Eight** presents a discussion of the research findings and offers an analysis of the general issues that have led this research. This chapter provides a theoretical discussion of the research findings and serves as the conclusion of this study.