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

DISCUSSION

This study describes the situation of genetic services in China and discusses the governance of genetic medical services and involves social and ethical issues, and it analyzes the roles that the agency of interest groups, such as hospitals, clinical geneticists, biotech companies, policy-makers, pregnant women, genetic disorder carriers and affected individuals and their families, play in the application of genetic testing. The aim of this study is to offer a way to understand the application of genetic testing in Chinese social, economic, political and cultural contexts from a social science perspective.

THE MAIN RESEARCH FINDINGS

In China, clinical genetic testing and genetic counselling are combined. Genetic counselling clinic is in charge of both counselling and testing service. The services are offered by clinical geneticists and are under regulation of official guidelines with the goal of reducing birth defects. Through a comparative approach in Chapter Three, this study points out that it is different social contexts that make genetic counselling distinctive. It argues that the economic conditions, non-professional genetic counselling provider, lack of full coverage of the healthcare system and national population policy are formative factors of the current mode of Chinese genetic counselling. Compared to the UK, where individual well-being and autonomous reproductive
choice are more considered in the governance of genetic testing, patients in China have fewer feasible options concerning reproductive decisions. Such situations contribute to the practice of genetic counselling and its governance in China, and, to some extent, limit proper communication and applicability of non-directiveness in genetic counselling in China.

Presently, prenatal genetic diagnosis is regulated by official regulations promulgated by the Ministry of Health, and qualifications are required to clinically develop prenatal genetic testing. However, there is no law or regulation specially regulating the application of predictive genetic testing, such as susceptibility (or predisposition) tests, and no permission from the Ministry of Health or the Ministry of Technology is needed. In Chapter Four this study shows that some biotech companies that had acquired business licenses have been carrying out predictive genetic testing as a technical business. In practice, there is no universal standard for describing genetic risk and no authoritatively approved criteria for test reports, which may cause the reliability of test reports to be in doubt. Chapter Four argues that, due to a lack of proper formal regulation, the commercialization of genetic testing has led to bioethical problems related to dubious advertising practices and misleading and unprofessional medical advice, of which the consequences for the users of tests are barely known. Biotech companies reap profits through predictive genetic testing, but test results only offer the level of potential risk probability, and no responsible provisions can be made for the potential patients.

In practice, the situation exits that employers and insurance companies ask for genetic information, or turn to predictive genetic tests into a requirement for employment and insurance, such as was the case in ‘the first genetic discrimination lawsuit case’ described in Chapter Four. Such a situation illustrates that, to some extent, not only personal autonomy and genetic privacy are at risk of being violated, but also discrimination occurs as a consequence of genetic testing. This makes ‘potential patients’, who have a predisposition to genetic disease or are carriers, vulnerable to discrimination and stigma in society. But, compared to the ‘potential patients’, it is clear that the patients with a genetic disease and their families are more vulnerable.

Chapters Five and Six examined thalassaemia and Duchenne muscular dystrophy (DMD) as case studies. Based on the practice of genetic counselling and testing for
thalassaemia and DMD, the research set out to illustrate the socio-cultural and economic conditions of the families with children affected by thalassaemia or DMD, especially shedding light on the difficult position of the mothers. They discuss related ethical and social issues and explain the vulnerability of those families. There are shortages of social welfare, social care and support for the patients. Besides the financial pressure caused by medical treatment, the families are also under heavy psychological pressure as a result of social stigma, as in some cases shown in the Chapters Five and Six. Families with thalassaemia suffer the heavy economic burden for lifelong blood transfusion for the affected children. Pre-implantation genetic diagnosis (PGD) and prenatal genetic testing make it technologically possible for prospective parents to create a donor match known as a ‘saviour sibling’. Although such a motive is ethically controversial and initiatives may have far-reaching consequences, the restriction of choice in some cases leads parents to decide to have a ‘saviour sibling’ by ignoring the potential risk of failing technology and failing to take seriously the process of informed consent when undergoing invasive clinical interventions during pregnancy. In practice, the families having children affected by DMD undergo economic hardships caused by the cost of ineffective treatment and the energy and time consumed in seeking assistance for their children. Additionally, Chinese cultural values attached to healthy offspring and traditional boy-preference increase the psychological burden of these families, especially for the mothers, who usually experience strong self-condemnation and are blamed by family members. This research indicated that the families affected by DMD are vulnerable, and the existing social discrimination and shortage of social care for the disabled aggravate their vulnerability.

Chapter Seven discusses of genetic services, eugenics and population quality. Research indicates that besides social, economic and cultural factors, political implications, such as the national population policy, social healthcare system and social welfare system, also play an important part in the formation of the practice of genetic testing and counselling and of the condition of the families affected by genetic disorders. Along with the implementation of the family planning policy, population quality and the notion of ‘well birth’ have been highlighted. The government has produced much propaganda related to improving population quality. In practice, prevention of and reducing the number of birth defects is the actual goal
of prenatal genetic diagnosis. With the eager wish to reduce birth defects, a eugenic approach was adopted as a legal means. These social and political contexts provided society with views that looked upon birth defects as a burden for both society and family, which may exacerbate genetic discrimination and social stigma and worsens the vulnerability of the individuals and families affected by genetic disorder.

THE SITUATION OF GENETIC TESTING SERVICE IN CHINA

Medical genetic service mainly includes genetic counselling, genetic diagnosis and prenatal genetic diagnosis and pre-implantation genetic diagnosis. Although clinical genetic counselling/testing started in China in the 1980s, a formal clinical genetic services system has not yet been established (Zhang 2008). Medical resources are insufficient and their nationwide distribution is unbalanced in China. Most of hospitals or healthcare providers, including many first-class hospitals, are not qualified to offer genetic counselling and testing. In some large hospitals, where a clinical genetic service is available, the test items are limited and comparatively costly, which is hard for most people to afford. It is fact that it is ‘costly to see a doctor, and more costly to see a doctor for genetic disease (kan bing nan, kan yichuan bing geng nan)’. In fact, it common that families fall into poverty because of having a genetically affected child, especially in rural areas. The vulnerabilities of such families has been argued in Chapters Five and Six through discussing the difficulties encountered in daily life by families affected by thalassaemia or DMD.

In China, genetics clinics and the clinical genetic services are few in number and lag behind demand. Hospitals or health providers do not have an independent genetic department, although some hospitals have a genetic counselling clinic and carry out clinical genetic services. As was described in Chapter Three, the genetic counselling clinics combined with genetic testing usually are organized under the department of paediatrics and/or department of obstetrics. Some well-known Chinese geneticists, believe that the reasons why there are fewer genetic clinics than needed is that the application of medical genetics such as genetic

45 In Chinese, upper-class hospital is called san ji jiadeng yiyuan. According to the administrative system for hospital, it is the top level of hospital.
testing based on advanced genetic technology requires investment to update equipment and to train the participants. Additionally, genetic testing is more challenging and has greater potential risks. Therefore, hospital administrators do not believe the clinics can be profitable and thus do not provide deserved support for the development of clinical genetics (Huang & Gao 2006).

In practice, although medical genetics have improved and have been contributing to the prevention of birth defects, as was shown in Chapter Seven, there is still no professional title for medical geneticists. The doctors working in medical genetics come from different fields. They have professional titles in those fields although they work in genetics. As the qualification of the medical practitioners is required and enforced, they have to take examinations on their original specialties, such as paediatrics, obstetrics, internal medicine and so on. It is difficult for them to keep up to date with advances in the fields they no longer practice. In order to pass their annual examinations, they have to spend time taking part in the clinic activities related to their original specialties rather than to medical genetics. This weakens their potential in medical genetics, and even turns them away from medical genetics. This also exacerbates the shortage of clinical genetic services. Additionally, regular professional training for clinical genetic service teams has not yet been shaped, which also adds to the shortage of qualified professional genetic personnel and the specialized skills of service teams. Furthermore, some hospitals suspend or stopped performing genetic testing, especially for prenatal genetic testing.

In practice, the laboratory result of prenatal genetic testing is not always reliable. There have been cases in which incorrect test results from prenatal genetic tests have resulted in lawsuits, which may make some hospitals hesitant and unwilling to perform prenatal genetic testing. In fact, besides the large financial burden of testing, a shortage of medical genetic services also makes it more difficult for patients to get access to these services.

However, contrasting with the shortage of prenatal genetic service, the commercial testing service offered by biotech companies is mushrooming. As demonstrated in Chapter Four, the emergence of commercialization of predictive genetic testing is stimulated by economic profits, and the fact is that it lacks adequate regulation. In return for a steep fee, the companies offer a report to the clients who undertake predictive genetic testing, with results
provided on the clients’ predisposition status and the offering of unprofessional medical
advices. To some extent, enticed by tempting advertising practices, undertaking commercial
genetic testing is a luxurious consumption by the wealthy members of society. For those
people who are in dire need of genetic services, such as people who have a family history of
genetic disorder or the pregnant women who are at high risk to have a affected fetus,
commercial genetic testing does not offer a testing service for prenatal diagnosis. Thus,
biotech companies cannot offer feasible and tangible services for those people who need it
most. Additionally, this kind of genetic service involves many social and ethical issues, as
was discussed in the Chapter Four.

As mentioned above, my study offered a way to understand the current practice of
genetic testing in China. It also reflected the fact that the availability of genetic medical
assistance for the individuals who really need it is far from satisfactory.

INFORMED CONSENT IN THEORY AND IN PRACTICE

The Council of International Organizations of Medical Sciences (CIOMS) guidelines give a
concise definition of informed consent: *A decision to participate in research made by a
competent individual who has received the necessary information, has adequately understood
the information, and after considering the information, has arrived at a decision without
having been subjected to coercion, undue influence, inducement or intimidation* (CIOMS
2002). The concept of informed consent is spreading globally and is discussed and addressed
by contemporary international and national bioethical guidelines. Informed consent is based
on the notion of individual autonomy. Autonomy is a basic principle of ethics and requires
that the individual’s right of self-determination be respected. Informed consent, it is believed,
ensures that patients can decide autonomously whether to permit or refuse actions that affect
them, and is treated as an essential part of any medical procedure. It has served as a
cornerstone for the development of the discipline of bioethics. Based on the importance of
autonomy in moral discourse, it has been argued that healthcare professionals are obligated to
engage patients in discussions regarding the goals and that the patients are the final
decision-makers regarding all therapeutic decisions. In the legal arena, informed consent has
been used to develop standards for doctor–patient interactions and clinical decision-making. It is widely accepted that legal standards require the doctor to inform patients of the risks, benefits, and alternatives of all proposed treatments and then allow the patient to choose among acceptable therapeutic alternatives (Berg et al. 1987).

In China, several laws, such as *Law of Torts, Law on Licensed Doctors and the Measures for the Administration of Prenatal Diagnosis Technology*, stipulate that patients have the right to know and, correspondingly, doctor have obligation to inform. Although such laws do not regulate what to inform and how to inform, theoretically doctor should inform the patient of whatever a reasonable physician would inform the patient under the circumstances. In fact, the stated intention of, and the importance attached to, informed consent is to protect the autonomy of patients by offering adequate information for patient to make voluntary decisions (Andanda 2005; Hansson 2005). However, my study found that in clinical practice medical geneticists understand the connotation of informed consent and implement the informed consent procedure in their own way.

Although there has been some support for informed consent within academic fields, there seems to be little enthusiasm for it in medical practice. My study shows that in clinical genetic practice, informed consent, to some extent, is used in a way which is contrary to its original intention. Doctors typically would think of informed consent as a legal requirement for a signed piece of paper rather than seeing informed consent as a process that promotes good communication and patient autonomy. Many doctors hold the view that informed consent is a complex and legally prescribed recitation of risks and benefits, and that most patients find the possible risks listed on the informed consent form frightening or confusing. In practice, what to inform and how the informed consent procedure is carried out do not get the attention they deserve. Generally, most patients are unable to understand well the professional language, medical terms, and the complex medical information in the consent form.

Chapter Three of this thesis shows that clinical geneticists complain that they do not have enough time or energy to develop a good communication with patients because of the large number of visitors to the clinic sessions. Under normal conditions, if the doctor requires patient to sign a consent form, patient will sign, or his/her proxy will sign instead, and
sometimes patients sign without carefully reading the form. The patients know it is required and they know they have to sign the consent forms because they cannot acquire medical treatment unless they do so. Chapters Five and Six, which take thalassaemia and DMD as case studies, examine the practice of genetic testing and informed consent in the clinical practice of genetic testing. As mentioned in Chapters Five and Six, without a signed informed consent form, clinical geneticists will not prescribe a test, especially for prenatal genetic testing. In clinical practice, doctors understand and implement the informed consent by standing on and defending their own position. On some occasions, informed consent is more likely to be used to avoid potential trouble and, to some extent, to shift responsibility of risks-taking onto the patient. The patient’s signature on a consent form can be treated as written evidence of a patients’ permission if a dispute should arise. Such a situation sometimes happens in the practice of genetic testing, as exemplified by the occurrence of failing prenatal genetic tests, as discussed in Chapter Five. This chapter discuss one case in which the prenatal genetic test failed to offer correct test results and another case in which the signed informed consent forms served as protections for unethical behaviour by test providers.

Theoretically, informed consent is expected to benefit patients. Academia also pays much attention to informed consent: not only professional ethics, but also law and regulation, define informed consent as a patient right. However, in clinical practice, it becomes clear that doctors and patients sometimes have different understandings of informed consent. And to some extent, the original intention of informed consent, which proceeded from the patient’s right to make a voluntary and free decision on the basis of awareness of all available information on the medical treatment, is still a long way from being achieved. This reflects the vulnerability of patients in such contexts.

**GENETIC TESTING, GENETIC DISCRIMINATION AND STIGMA**

Genetic testing is often used as a form of genetic diagnosis to indicate the existence of a genetic disorder. The identification of a disorder does not necessarily lead to treatment options, because there is currently no effective intervention or treatment available for many
genetic conditions. Sometimes the identification of a serious genetic disease is the cause of social stigma. Such stigma may derive from the widespread view of religions retribution as an explanation of pathogenesis. These social conditions were discussed in Chapter Six. Often, genetic testing is used to identify carrier status. Carriers are persons who are themselves unaffected but who are at risk of having affected children. If carrier status is confirmed, the carriers can be aware of the risk to give birth of an affected child. Prenatal genetic testing and subsequent selective abortion of an affected fetus could be chosen. This study shows that in China this is a formally accepted way to prevent a genetic disorder, and it was also an encouraged method to reduce birth defects due to serious genetic disorders, and therefore to improve the population quality. Such conditions are discussed in Chapter Seven of this thesis. Additionally, my study also shows that the information of carrier status sometimes causes genetic discrimination. If predictive testing was inappropriately used and the information of carrier status or predisposition was disclosed, the test results might cause unjust genetic discrimination. My study also showed that in China there was growing concern that genetic information might be used in nonclinical contexts, which may harm the interests of the individual or his/her relatives. One example was the case study on genetic discrimination, in Chapter Four, described as ‘the first’ case of genetic discrimination in China. The discriminator in this case was a government institution, which officially spread the opinion that persons who carried a certain genetic disorder gene were unwelcome in society. In fact, there have already been similar cases in which thalassaemia carriers had experienced employment discrimination. People lost their chance to get a job simply because of their carrier status. In Chapter Five, this study also mentioned that the parents of children affected by thalassaemia would rather hide the condition of the child, treating it as a family secret. The families were afraid that they and their children would be discriminated against and ostracized.

The commercialization of genetic testing might have made it easier to gain access to genetic testing for people outside the public health sector. As shown in Chapter Four, commercial genetic testing is still unregulated, which may worsen forms of genetic discrimination. In United States during the 1970s, the genetic discrimination against the carriers of sickle-cell disease led several states to enact legislation that specially forbade the
use of sickle-cell testing to determine insurability and employability (Reilly 1975). In China, when the first case of genetic discrimination was reported, it caused debate, and some experts appealed for the legislation on genetic antidiscrimination. But, as my thesis discusses regarding the relationship between the ‘superior birth’ and genetic discrimination in the section entitled Younsheng idea and genetic discrimination in Chapter Seven, the ‘superior birth’ policy, especially the eugenic approach adopted to reduce birth defects in China, still has a strong influence on the people and society. Perhaps this was one of the original bases of genetic discrimination. Thus, we have to say that there might be a long way to go to achieving legislation on genetic antidiscrimination in Chinese contexts.

At the same time, the identification of carriers causes psychological harm to the carriers, especially for the mothers/parents who passed down the disorder to their children. Usually persons suffering from a severe inherited disease and their close family are socially stigmatized, as shown in the cases study in Chapters Five and Six.

For young unmarried carriers, the identification of carrier status perhaps stigmatized them and gave them an inferiority complex. In fact, it also made it difficult for them to find a partner and put them under a heavy psychological burden due to the dilemma of whether to disclose the carrier status to their likely partner. It was more difficult for the X-linked disorder carriers who are female. Additionally, the traditional Chinese gender preference and the cultural value attached to the male offspring in a family sharpened the stigma of the families with a boy with DMD, as shown in Chapter Six. However, under the circumstances in which improving population quality is the aim, doctors usually proceed from the idea of the prevention of genetic disease. They would consider the recommendation of an available test to be in the patient’s best interest. Some local governments launched campaigns for free carrier testing, as described in Chapter Five in the context of carrier testing for thalassaemia. In such a context, carrier testing is very much encouraged. But the discrimination and stigma that might be caused by status identification have not yet received the attention deserved.

CHOICE AND DECISION-MAKING

In practice, the choices of taking a prenatal genetic test concerns reproductive decisions,
which are highly personal, and the choice of being tested and the decision-making should be autonomous and based on the considerations of personal benefit and personal preference. In theory, before undertaking genetic test counselling, the counselee should know the possible disadvantages and advantages of undertaking the test. Theoretically, genetic counselling is considered traditionally non-directive – that is, counselling provides sufficient information to allow families or individual persons to determine their best course of action for themselves, and avoids making testing recommendations (Michie et al. 1999). Non-directiveness embodies autonomy, one of the recognized principles of medical ethics. But in practice, non-directiveness is difficult to achieve, even in those countries such as the United Kingdom, which has genetic counsellors with professional education. In practice, the counsellors and clinical geneticists usually regard their counselling as non-directiveness, and state that their counselling is non-directive (see Chapter Three). In fact, all doctors desire their medical activities to be regarded as ethical. There are good reasons why clinical geneticists and genetic counsellors might wish to claim that their work is non-directive. The declaration of non-directiveness is in keeping with the ethical requirement of autonomy.

As some scholars point out, a stance is considered to protect the doctor from any confusion with, and moral contamination from, the eugenics movement, and this will be useful to genetic counsellors both in public debate and internally within themselves. It protects the counsellors from overinvolvement with clients and perhaps also from litigation (Clarke 1991; Clarke 1997; Schneider 1998). Although non-directive counselling more respect personal autonomy, in some instances directive recommendation is also needed. For example, if the information of genetic testing results is also in the interests of other family members, such information may need to be disclosed to help them to know about their risk of developing a genetic disorder or of having a child affected by a genetic disorder. In this situation, considering the interests of involved family members, perhaps the counsellor or doctor needs to attempt to persuade the client to disclose personal genetic information to other family members and give directive advice on disclosure, which in theory is call an ethical recommendation (Gray & Clarke 2000). Usually, non-directiveness in counselling has close connection with the autonomous decision-making. In the context of prenatal genetic testing, non-directive counselling meets the ethical requirement that the prospective parents
have a right to make autonomous choices and decisions regarding their reproductive action.

However, my study found that in China the choices for prospective parents who are at risk to have an affected child were limited. Without socialized medicine, lack of social welfare and support for the disable and the sick, the guidance of Chinese population policy, potential social stigma and discrimination, all these social factors limit the options for the prospective parents to make reproductive decisions. In current clinical practice in China, non-directive genetic counselling, to some extent, cannot offer more effective and realizable ‘freedom’ or ‘autonomy’ to help and support patients’ decision-making. In fact, under the social factors in the Chinese contexts, it is difficult or impossible to achieve ‘autonomy’ in reproductive decision-making. As I discuss in Chapter Three, with the goal of preventing genetic disorder through reducing birth defects, it is difficult to achieve non-directiveness in counselling or advice from clinical geneticists. In practice, ‘free’ reproductive decision-making is not free or autonomous because limited options, and in many occasions it is actually a choice of ‘only one option’ or a choice of ‘no choice’.

EUGENICS AND BIRTH DEFECTS

From the research on the current situation of genetic services, this thesis shows that in China, genetic services of premarital, pregestation and prenatal genetic screening, genetic counselling and testing, generally, have the same fundamental and ultimate goals, that is, to avoid the recurrence of a particular genetic disorder, and to prevent and reduce birth defects. It shows the roles played by the genetic services in the prevention of birth defects and in the improvement of population quality in China. As described in Chapter Seven, clinical genetic services, especial the application of genetic counselling and testing in China, have reached the outcomes in prevention of birth defects and in a reduction in the number of children affected with certain genetic disorders by the selective abortion of affected fetuses. In Chapter There and Chapter Seven, my study shows that the current practice of genetic counselling in China is a medical-preventive model that attempts to avoid the recurrence of a particular genetic disorder and with the aim of optimizing population health through the prevention of birth defects. Chapters Five and Chapter Six, through the case studies of
thalassaemia and DMD, also argue that genetic services in China has played an important role in enhancing couples’ awareness of the prevention of birth defects.

This thesis has also argues that, in China, there are many factors, especially related to economical conditions, the national healthcare system and the population policy, that have limited the reproductive choices for parents. Hence, in Chinese social contexts such genetic services did not promote individual decision-making and autonomy, and in practice these are not regarded as successful outcomes of the genetic services. The thesis also analyzes the relationship between eugenics, Chinese eugenics [yousheng], genetic services and the prevention of birth defects in China. The term eugenics derives from the Greek word for ‘well born’. The Chinese word ‘good birth’ [yousheng] has the same meaning as the Greek word ‘eugenic’. It is also consistent with Galton’s core eugenics doctrine of improving the stock of humankind by the application of the science of human heredity. In this sense, Chinese yousheng could be translated to ‘eugenics’. Chinese eugenics is different from the ‘eugenics’ movement of the 1920s, but during the nationwide carrying out yousheng policy and efforts to prevent birth defects, the eugenic approach had been inappropriately accepted. Such an approach aimed to reduce birth defects through coercive prohibition or limitation of reproduction of people without ‘reproductive value’. Chinese scientists claimed that Chinese eugenics was not motivated by racism. Chinese yousheng is ‘eugenics’ in name only, and that, in practice, it is ‘prevention of birth defects’ and it does not go as far as would be expected of a eugenic movement (Ruan 2002). However, a eugenic approach that accepts the compulsory reduction of birth defects displays tendency to the eugenic movement. Chapter Seven discuss the eugenic approach, which has been abolished, and points out that although the ‘eugenics’ in local regulation have been annulled, its implications cannot be changed in a short time, and, to some extent, continues to aggravate genetic discrimination and stigma against the mentally retarded and the handicapped.

**VULNERABILITY**

With advances in genetic technology, its practical applications play an important role in prevention of genetic disease and the reduction of birth defects, and under the direction of
national population policy, to some extent, genetic services have been treated as an effective way to actualize improving national population quality. Chapter Seven of this thesis shows such conditions in China. Additionally, this thesis is concerned with the way in which genetic information may be abused for discrimination, particularly against the people who are affected by a certain genetic disorder, or at increased genetic risk of developing certain disorders. In fact, through describing the situation of genetic testing practice and discussion on the involved social and ethical issues, almost all the chapters of this thesis concern the vulnerable situation of the patients with genetic disease and their families. To some extent, this thesis offers an insight to understand the vulnerability of the individuals who are sufferers or carriers and their families in Chinese contexts.

The vulnerable situation of the family at a social level

Considering the situations of families with an affected child as shown in Chapters Five and Six, in China the families with genetically affected children were in a difficult situation in society. It is a fact that the families fall into poverty because of having a child with a genetic disease, or fall back into poverty even though they had already extricated themselves from poverty [tuo pin]. Poverty caused by disease is not unusual, particularly in the countryside of China, where many families cannot afford the expense of treatment and then have to give up treatment for their children. In fact, parents are under a heavy psychological strain when there is not enough money for medical treatment for their children. Chapter Five gives examples of families with children affected by thalassaemia. In fact, due to lack of organized and accessible social support and help, most of families face financial difficulty and take a long time to overcome such burdens. Besides the economic disadvantages, the affected individuals and their families also encountered social blame, stigma and discrimination. The application of prenatal genetic testing, together with the selective termination of affected pregnancies, has made society less tolerant towards serious congenital birth defects. In practice, this leads to social blaming of parents who do not undergo genetic tests and subsequently gave birth to a child with a disability, although sometimes they have no access at all to knowing their risk. At the same time, the disability and difference caused by genetic disorders results in discrimination in enrolment, employment and marriage. Also, the traditional social
interpretation of genetic disease seriously stigmatizes families. Chapter Six discusses such stigma by taking families with children affected by DMD as examples. Besides these, nationally social circumstances virtually impressed and transferred the negative messages that may engender stigma and discrimination on the carriers, the affected individuals and their families, such as the conducting of propaganda on ‘good or superior birth’, ‘improve population quality’, especially the continued influence of legal ‘eugenic approach’ adopted to reduce birth defects, although it had already been abolished.

Additionally, the government’s or the authority’s estimation of the ‘social burden’ caused by birth defect with genetic disorders actually passes on the impression to the public that congenital physical disabilities and mental retardation are a ‘social burden’ to the country. Parents and families might be blamed by society for adding to the social burden. To some extent, such information virtually may cause negative views and may sharpen the stigma and discrimination against affected individuals and their families, and then aggravate their vulnerability.

The vulnerable situation of the female at the family level

In society, genetically affected individuals, genetic disorder carriers, person with a high risk of developing genetic disorders, and their families are in a vulnerable position. Beside the financial and psychological vulnerabilities caused by society, some individuals are also under heavy pressure within the family. Money difficulties, stigma and discrimination from society may make parents blame each other, which makes the family relationship tense and strained. Additionally, females are more vulnerable within a family. From the case studies in Chapters Five and Six, it can be seen that the mothers of affected children were all under the psychological burden of self-condemnation. In the Chinese traditional family culture, it has long been widely accepted that the taking of a wife is mainly to carry on the family line, so the wife in a family naturally has a reproductive duty and is expected to give birth to offspring, particularly of son. Having an abnormal child makes the wife feel guilty and herself to be inferior, and in some cases results in being blamed and abominated, and even divorced, by her husband, especially in X-linked disorders such as DMD. The blame of family member creates guilt and psychological pain. In Chapter Six, the study describes the
very vulnerable condition of the children with DMD and their mothers. Although, in practice, prenatal genetic testing offers a possible way for the parent to terminate an affected pregnancy to avoid an affected birth, it also brings heavy pain and suffering of mind and body for the mother. The study also discloses that such sufferings of a mother seem often to be ignored due to the eager desire for ‘good birth’ and strong wish to exterminate birth defects, which reflect the weaker and more vulnerable position of the mother in a family, and thus in society.

From a social science perspective, this study describes the practice of genetic testing and analyzes the social roles of social factors, such as Chinese social economy, national policies and culture, played in its formation and application. Through discussion of such factors that shaped the views and actions regarding reproductive decision-making of the involved population, and sharpened the reproductive pressure, especially on females, and through discussion of the involved social and ethical issues, the study identifies vulnerable populations with respect to the practice of genetic services in China, and it delineates the social factors that influence, shape and sharpen their vulnerability. Thus, the study achieves its research objective. It offers an authentic and a diaphanous insight to understand the situation of vulnerable populations in the genome era with the quick development of genetic technology in the Chinese socio-economic and socio-political contexts.