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

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Citation for published version (APA):

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CHAPTER TWO

BRIEF OF RESEARCH CONTEXT

(Interview a couple who has a thalassaemia affected child)
THE DEVELOPMENT OF MEDICAL GENETICS IN CHINA

This study is based on the application of medical genetic knowledge and the techniques which have been developed to this end during the past decades in China. I open this chapter by looking back at the development of medical genetics in China.

The study of genetics in China started at the beginning of the twentieth century when Chinese scholars began their work on human genetics by surveying the frequency of the ABO blood group among the national population. The results were published in 1918, and this was the first study of gene frequency among the Chinese. In 1922, Chen Zhen, one of China’s genetics pioneers, opened genetics courses in the National Southeast University. In 1937, reports on inherited diseases among the Chinese, such as on the frequency of colour blindness, were published. In 1948, Li Chingchun, a famous geneticist, wrote the *Introduction to Population Genetics*, which became a well known textbook in the field of genetics and from which a whole generation of geneticists benefited (Huang & Gao 2006).

However, the development of genetics in China was not always plain sailing. It fell into difficulties in the 1950s and during the ‘Cultural Revolution’. Chinese geneticists faced the suppression that came from the Lysenkoism, which was spread from the Soviet Union to China in the early 1950s. Those Chinese scientists who believed in Mendelism-Morganism were suppressed by heavy political pressure, and their teaching and research on genetics came to a total halt. Some geneticists were forced to give up their research projects or to leave China.

The situation began to change with the introduction of the policy entitled ‘let a hundred flowers blossom and a hundred schools of thought contend’ (baihua qifang, baijia zhengming), which commenced in 1956. In August of that year, the Chinese Academy of Sciences (CAS) and the Department of Higher Education of Ministry of Education (MOE) co-convened a symposium on genetics in Qingdao, Shandong province. The symposium was a significant and historic turning point in the development of Chinese genetics (Tan & Zhao 2002), considered both a very successful academic meeting and also as a meeting of
‘rehabilitating’\(^1\) for those geneticists who believe in Morganism. After this genetics symposium, courses on genetics were reopened in colleges, the genetics scientists returned to their positions, and their research work restarted. For example, the Beijing Agricultural University founded two genetics teaching and research groups, one of which is for research on Morganian genetics, and another on Michurinian genetics (Tan & Zhao 2002).

However, before ten years had passed, the ‘Cultural Revolution’ started in 1966. This blocked scientific development again. Genetic research was suspended until 1978, when the central government decided to carry out democratic reforms, and genetics was resuscitated and received comprehensive development. Several associations or societies have been established since 1978 to match the development of medical genetics, such as the Chinese Society of Genetics and the Association of Medical Genetics. With the leadership of these organisations, genetic research, education, and clinical services developed rapidly. In 1978, departments of medical genetics were established in medical colleges such as Sun Yat-sen Medical University, Hunan Medical University, and Peking Union Medical College. In the same year, prenatal genetic diagnosis was carried out for the first time. Following this, the Peking Xiehe Hospital started offering prenatal genetic diagnosis for thalassaemia by karyotyping foetal cells cultured from amniotic fluid with technical support from the Chinese Academy of Sciences.

Genetic counselling clinics, where paediatricians or obstetricians offered counselling, were opened in the early 1980s. In 1981, prenatal diagnosis could be done in the first trimester to detect the X chromatin of the foetus through testing chorionic villi collected at the seventh or eighth week of gestation. During the 1980s, genetic technology began to develop rapidly, and in the 1990s genetic diagnosis could be performed on both amniotic fluid cells and chorionic villi. Genetic analyses of phenylketonuria (PKU), haemophilia, Duchenne muscular dystrophy (DMD), glucose-6-phosphate dehydrogenase (G6PD) deficiency, fragile X syndrome, and Huntington’s disease, amongst others, were performed, and such prenatal diagnosis became available in some of the larger hospitals (Huang & Gao 2006). At the present time, on the national level some officially qualified hospitals are

\(^1\) In Chinese, ‘rehabilitating’ is visually expressed as *zhai maozi*, which means ‘removing hat’, and literally means removing an incorrect rating.
offering chromosome analysis on foetal cells, and more than twenty genetic disorders can be diagnosed.

RESEARCH DOMAIN

With the development of genetic technology, the interplay between its practical application and the relevant political, economical, and cultural characteristics in a given society is becoming more and more noteworthy. Much research has been done in the socio-genomics field. For example, Kaushik Sunder Rajan, in his book *Biocapital* published in 2006, illuminates how the contemporary world is shaped by the marriage of biotechnology and market forces. Nikolas Rose also points to how a new kind of citizenship, which he terms ‘biological citizenship’, is taking shape in the age of biomedicine, biotechnology, and genomics. Rose’s book, *The Politics of Life Itself* published in 2007, offers an examination of recent developments in the life sciences and biomedicine that have led to the politicisation of medicine, human life, and biotechnology. Reyna Rapp’s book, *Testing Women Testing the Fetus* published in 2000, illuminates the social impact of amniocentesis in the United States. Her work provides an incisive account of the realities of stratified reproduction by describing the scientific and the social practices of amniocentesis. Rapp’s study shows how people weave genetic knowledge together with their concepts of parenthood, childhood, family, and work, and documents the complexity of the social, cultural, and technological circumstances surrounding genetic testing. Further, Sarah Franklin and Celia Roberts’ book, *Born and Made* published in 2006, examines the case of pre-implantation genetic diagnosis (PGD), the procedure used to prevent serious genetic disease by embryo selection and result in the so called ‘designer baby’. *Born and Made* provides a deep sociological examination of the ethical challenges and competing moral obligations that define the experience of PGD.

These studies are significant in the research filed of genomics from a social science perspective. The studies indicate the issues emerging under the impact of the application of genetic technology. In fact, genomics can only be understood well in relation to the social contexts where it emerges. Such studies are most often based, however, on developed western countries such as America or the United Kingdom, countries which have a leading scientific
role in genetic technology. By now few studies, in English or in Chinese, exist which examine the repercussions of developments in genomics from a social science perspective for Chinese society, and no social science studies focus on genetic testing in China.

My research is empirical and interdisciplinary. It is based on empirical fieldwork conducted in China and focuses on the practical application of genetic testing. The research questions all arise from Chinese socio-economic, political, and cultural contexts. My research aims to describe the developments in a society which is more stratified, from the perspective of people who are vulnerable in terms of poverty and human rights. My research offers a special perspective in studying the social aspect of genomics for Chinese society.

According to the provider and the purpose of genetic testing, my research presents the practice of genetic testing in China from three angles:

One: Genetic testing offered by biotechnology companies. In China, biotechnology companies offer predictive genetic testing that aims to detect susceptibility for developing certain multi-genetic diseases. In my research, I name this kind of genetic testing commercial genetic testing. The commercialisation of predictive genetic testing and the involved social and bioethical problems are presented in Chapter Four.

Two: Genetic testing offered by hospitals. The testing to diagnose genetic disorders in children or foetuses and to detect carriers, combined with the service of genetic counselling, is performed in hospital obstetrics and/or paediatric clinics, which is mostly referred to as clinical genetic services in this research. In qualified hospitals which have received special permission to offer prenatal diagnosis, specialist doctors who have been educated in genetics but not specially trained in counselling perform genetic counselling and tests. Such clinical genetic testing practice is medical and the counselling is not non-directive as is the norm in the western – and global – guidelines. The practice of genetic counselling and analysis and the issues concerned are described in Chapter Three, by way of a comparison with professional genetic counselling in the UK. The testing done in hospital clinics and the vulnerable condition of individuals are described in Chapters Five and Six, based on the case study on Thalassemia and DMD.

Three: Testing as part of a preventive eugenics policy. Under the population policy that aims to reduce quantity and improve quality, testing offered for premarital examination,
prenatal diagnosis, and newborn screening play an important role in the prevention of birth defects. The general situation of genetic services, their effect on national eugenics policy, and the social implications in the Chinese context are presented in Chapter Seven.

FIELDWORK SITES, INTERVIEWEE SELECTION, AND DATA COLLECTION

Fieldwork for my research was conducted mainly in mainland China from 2006 to 2009. In total, four phases of fieldwork were carried out: from July to September 2006, from February to May 2007, from September 2007 to March 2008, and from September 2008 to February 2009 (September to November 2008 in China, December 2008 to February 2009 in the UK). Considering the diversity of the natural and cultural environment, social and economic conditions, health resource distribution, and regional prevalence of thalassaemia, different places were chosen for field research. Among the fieldwork sites were hospitals, maternity clinics, patient organisations, biotechnology companies that offer genetic tests, and research institutions and universities that are researching genetic technology. Each period of fieldwork had as the main task data collection for one part of the above-mentioned three parts of the research, but at the same time also allowing for information collection for the other parts of the research and updating previously collected information.

In the first phases of fieldwork that were carried out from July to September 2006 in Beijing and Shanghai, the main task was to understand commercial genetic testing in China. Interviews were the main method of data collection. The task was to understand the commercialisation of predictive genetic testing offered by biotechnology companies in China, and its social and ethical problems. During fieldwork, I interviewed agents of biotechnology companies, a geneticist, insurance agents, clinical doctors, and medical students, amongst others. I found, however, that it was not easy to make fruitful interviews with company agents, and sometimes I needed to develop strategies for creating opportunities to talk in more detail with agents; for example, by contacting them as if I was a common customer planning to take the test. In this way I contacted about ten companies and interviewed three agents.
The second fieldwork period was conducted in Beijing, Guangxi, and Sichuan from February to May 2007. During this period, through the participant observation in genetics clinics and interviews with clinical geneticists, I became acquainted with the application of prenatal genetic testing in China. The questionnaires and interviews conducted helped me to understand public opinion on prenatal genetic testing. Through previously established connections I had the chance to conduct observations in the ‘Home of Thalassemia’ in 303 Hospital in Nanning, Guangxi province, where I met many children with thalassemia and talked with them and their parents. I also made contact with some children with thalassemia through the media, such as newspaper or Internet reports.

Here I would like to give one example of how I sought and recruited interviewees. I read a court case report about genetic testing for thalassemia, in which was mentioned the name of a lawyers’ office that offered legal aid to the family in question. By contacting this office I located the lawyer who acted as the attorney for the family. Through this lawyer I obtained the contact details of the family and spoke with the parent. Later, this family introduced me to several families with thalassemia affected children. In this fieldwork, from around thirty children that I met or contacted, I chose eight families to interview in-depth and to follow up. Through the in-depth interviews with families that have children with thalassaemia, I gained firsthand information which helped me to understand the reproductive decision-making of thalassaemia carriers, their decision to produce a ‘saviour sibling’, and the social factors affecting their decision-making.

On the basis of the findings from the second fieldwork period, further fieldwork was conducted from September 2007 to March 2008 where I continued to participate in clinical genetic counselling sessions to understand the practice of genetic counselling, this time focused on counselling related to Duchenne muscular dystrophy (DMD). I was mainly situated in the genetic counselling clinic of Xiehe hospital. I attended nearly every session, two sessions one week, for two months, and through clinical participation I found many families from all over the country with boys affected by DMD. Finally, I chose three families

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2 The Home of Thalassemia in Chinese is *di pin zhi jia*. It is an outpatient department of 303 Hospital that specialises in offering blood transfusion treatment for children with thalassemia. 303 is the name of the hospital, which is a military facility.
as my primary respondents for the case study on DMD. By using in-depth interviews and observations of the families, their vulnerability and the difficulties they faced were well understood. This fieldwork period was the empirical basis for my case study on DMD, and it also offered basic information about the practice of genetic counselling in China.

From September 2008 to February 2009, international fieldwork was carried out in China, Hong Kong, and the UK. In-depth interviews were the main method used in this period, and clinical observation was conducted when possible. The aim of this fieldwork was to better understand the current application of genetic counselling in China by comparison with the UK. Before this period, the research conducted in China had collected information about the general situation of genetic counselling. During this fourth fieldwork period, through in-depth interviews with genetic counsellors and clinical geneticists, I acquired detailed information about how genetic counselling is carried out in practice, how the related social and ethical issues are handled, and the respondents’ opinions on such issues. Meanwhile, during the fieldwork conducted in the UK, following an introduction from the Bios research centre of the London School of Economic and Political Science (LSE), I visited four hospitals’ genetics departments and interviewed one clinical geneticist and eight genetic counsellors. I visited the United Kingdom Thalassaemia Society (UKTS), a charity institution for thalassaemia patients, and conducted an in-depth interview with its coordinator. Further, I conducted an interview with an executive member of Duchenne Action, an institution for patients with DMD. The latter interviewee was also the father of a boy affected by DMD. Such interviews helped me to understand more the difficulties of families affected by thalassaemia and DMD in the Chinese context, which was helpful for my related case studies.

The time frame and the main research methods used in each fieldwork period are presented in the introductory sections of Chapters Three to Seven. The selection of specific sites of fieldwork and participants for in-depth interviews was based on the consideration of the demand and objectives of each part of the research. It also depended on the information acquired through Internet searching or archival research as to what kind or which institutions might be suitable research targets. Usually, this was also the way I found potential interviewees, such as agents of biotechnology companies, agents of insurance companies, and the executive coordinator of patient organisations. Most often I gained access to interview
genetic counsellors, clinical geneticists, genetic researchers, and a haematologist through introductions from previous interviewees (the snowball effect). For example, sometimes my interviewees recommended other persons whom they thought might be suitable for me to talk with. My five years’ experience as a teacher in a well known medical university in Beijing also enabled me to build on some contacts with medical researchers and clinical doctors, which helped me to gain access to conduct participant observation in the clinics. Additionally, clinical observation and participation facilitated access to the pregnant women, patients, and their families for questionnaires and interviews, and the recommendation of other patients was also a way to recruit new patient interviewees.

Most of my interviewees in China were Chinese and did not speak English, with the exception of some doctors and clinical geneticists who had published academic papers in English. Therefore, Mandarin (putong hua, the officially normative Chinese) was the language used in most interviews. The interviews with genetic counsellors and clinical geneticists in Hong Kong were conducted in Chinese and/or English. For the interviews in the UK – eight interviews with genetic counsellors, one with a clinical geneticist, and three with the people from patient organisations – English was the language used.

The semi-structured interview was the main method of my field research. The outlines and interview questions varied according to the different target groups and informants. In-depth interviews were conducted by appointment. Each in-depth interview was unique, and the outline for an appointed interviewee was also specially designed. Some interviews with patients took their own course, as the respondent preferred to talk more about his/her views rather than to simply give answers to multiple choice questions; thus, the questionnaire would turn into an informal interview. In this way, some questions in the questionnaire were also used as interview questions.

Through such informal interviews with patients, gradually the cases for the case study took shape, and were selected for further exploration. Then the contacts were built up and the follow-up interviews and visits conducted. Telephone interviews were also conducted when an on-site interview was not possible. In my experience, an arranged telephone interview with good preparation could sometimes work very well. Usually, in order to encourage interviewees to talk more, the interview questions were open ended. Before the start of each
interview, usually I simply introduced myself and explained my research to the interviewee. The interviewees were informed that the interviews were just for this research, they agreed to voluntarily talk with me and answer my questions, they understood that the interview might be used in academic papers which possibly would be published, that their privacy and confidentiality were guaranteed, and that the interview content would be anonymous and their names and any identifying details would not be disclosed in any subsequent papers. The interviewees were also informed that they could feel free not to answer some questions, not talk about certain topics, or stop the conversion at any time. The interviews were semi-structured and started with easygoing questions. Regarding the research themes, there was seldom a limit placed on the discussion. During interviews, the conversation sometimes explored random topics and unexpected or unplanned themes, which offered more background regarding the social contexts. For the in-depth interviews, I asked permission from the interviewees to record the conversations. Under the terms of this permission, the interviews were recorded by digital recorder or MP3. Some phone interviews were also recorded, with the permission of the interviewees. Notes were taken during all the interviews at the same time as the conversation. All the audio files of interviews were saved on computer and/or USB disk, and other original data collected during the fieldwork were also kept by me. These data have been used solely for this research.