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

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CHAPTER SEVEN

GENETIC SERVICE, INTERVENTION RELATED TO BIRTH DEFECTS AND POPULATION QUALITY IN CHINA

(The course on family planning and good birth)

39 This chapter is under review with the international peer-reviewed journal: Journal of Contemporary China.
This chapter introduces the understanding of eugenics and Chinese yousheng, and their relationship to genetic services and the prevention of birth defects in China. It analyzes the important roles played by the genetic services of premarital, pregestation and prenatal genetic screening, genetic counselling and testing in the prevention of birth defects and in the improvement of population quality in China. The chapter points out that, as a national population policy, in China eugenics is not only a genetic term, but also has a certain social and political sense. The chapter argues that avoiding having a baby with a birth defect allows families and individuals to have easier life and also reduces financial burdens. However, given the national desire to reduce birth defects, some individual rights and choices involving personal genetic information and decision-making do not get the attention deserved. An insufficiency of psychological support and humanistic care in genetic services still exists, and the increased financial burden for the family and society caused by people with birth defects puts increased pressure on women and may sharpen genetic discrimination.
INTRODUCTION

In China, according to population policy and the Chinese Population and Family Planning Law, controlling population quantity and improving population quality is the purpose of family planning. With the effort to control the population quantity, improving quality of birth has attracted great importance (Ding & Hu 2000). Family planning policy has created a social environment in which parents themselves are keen to produce ‘superior’ offspring (Dikotter 1998). Under such policy, having a healthy baby is the minimum expectation for the prospective parents. Ensuring that the only child born to a couple is healthy and intelligent has become a common concern. Spreading the knowledge of genetics, generalizing the genetic services and adopting practical measures to avoid birth defects [chusheng quexian] have assumed increased significance. In practice, the prevention of birth defects is becoming increasingly important to government, the public and individuals. In recent decades, genomic research quickly developed, and the clinical application of genetics has been seen to hold enormous promise in the quest to identify the bases for inherited disease and to develop effective means of prevention (Petersen 1999). For persons at higher risk of producing a child with a birth defect, such as a person with a family history of a genetic disorder, or the prospective parents with the maternal age over 35, prenatal diagnosis and subsequent selective abortion are available and practicable for avoiding the birth of an affected child. In China, during the implementation of national population policy, the term of eugenics [yousheng] has been very broadly used and has become very well known, and it has become popularized and accepted. At the same time, genetic services and technology have rapidly developed in China. For a better understanding of the condition of clinical genetic service and opinions about yousheng of individuals, I conducted a survey in China during 2007 to 2008. I visited different levels of maternity hospitals and genetic counselling clinics and sat in on counselling sessions. Information and data were collected through interviews and my personal experience during the survey, which offer a way to understand the status of genetic services and the prevention of birth defects in China. This chapter analyzes the roles that the practical genetic services such as genetic screening, genetic counselling and testing play in
the prevention of birth defects and in the improvement of population quality, and it analyzes the issues involved in a Chinese context.

THE UNDERSTANDING OF ‘EUGENICS’ AND CHINESE YOUSHENG

The term eugenics derives from the Greek word for ‘well born’. It was first used in 1883 by Francis Galton, who described it as ‘the science of improving stock, not only by judicious mating, but by whatever tends to give the more suitable races or strains of blood a better chance of prevailing over the less suitable than they otherwise would have had’ (Galton 1883). His research, enhanced by statistical methods developed as he needed them, convinced him that society’s stock of talent could be greatly enlarged if members of favoured families were to increase their rate of childbearing, which is called positive eugenics. The balance should be further improved, he believed, by discouraging from reproducing those who had less to offer, which is called negative eugenics (Wikler 1999). Galton himself held that ‘the possibility of improving the race of a nation depends on the power of increasing the productivity of the best stock. This is far more important than that of repressing the productivity of the worst’ (Galton 1909). He launched a eugenics movement to improve the human race, or at least to halt its perceived decline, through selective breeding. His ideas were spread quickly, and by the 1920s eugenics movements existed in many countries. The lowest point in the history of eugenics was reached in Nazi Germany in 1933, where compulsory sterilization of those with mental illness or impairment, along with people suffering from various disorders, was followed by the killing of those individuals, together with those with congenital malformations. Eugenics was embraced by Hitler and his Nazi followers, tarnishing its name. Thus, eugenic ideas lost their cachet, becoming a largely discredited field and the term ‘eugenic’ now is used as a pejorative epithet (Wikler 1999). In China in 1920s, eugenics was introduced from Britain. Ting Wen-chiang [Ding Wenjiang], the well-known geologist, sociologist and anthropologist, translated eugenics as ze si (in Chinese, ze means wise and si means offspring) and used the term zesixue. In zesixue, Galton’s eugenic theory was introduced. Later, during the 1930s, the pre-eminent sociologist,
educationalist and eugenicist Pan Guangdan published several works on eugenics to disseminate Galton’s eugenics and claims about birth control, limitation of population quantity and prohibition of intermarriage, and pointed out the disadvantage of consanguineous marriages (Zhang 2001). From the foundation of the People’s Republic of China (PRC) until 1979, eugenics was treated as a ‘pseudoscience’ [wei kexue] and was simply linked to the sin of fascist racial slaughter, and was considered a forbidden area [jin qu] (Ruan 2002). In 1979, eugenics was advocated again. It was argued that not all the eugenicists were racists and that eugenics had its rationality with respect to improving population quality (Ruan 1980).

In recent decades, accompanying the enforcement of population policy and the development of genetics, eugenics has been popularized in China. And the progress in reproductive technologies and genetic engineering is also seen as a helpful step towards implementation of positive eugenics in order to improve the genetic quality of mankind and to produce ‘ideal’ babies. Calls for positive eugenics, designed to make ‘superior’ people breed ‘superior’ offspring, occasionally appear in the literature. Some scholars uphold such a view: ‘there is a clause allowing the parents of a first child who is handicapped to bear a second child: this should be replaced by a clause forbidding parents of a first child who has a hereditary handicap to bear a second child. At the same time, some people distantly related by blood with high intelligence quotients, such as outstanding scientists, mathematicians, writers, musicians, top sportsmen and others, should be encourage to have a second child within the family plan’ (Zhu 1990).

In 1993, the Minister of Health enacted the Chinese Maternal and Infant Health Law, which primarily was titled Yousheng Law, and was translated into English as Eugenic Law. The law mandates a premarital physical check-up for serious genetic diseases, some infectious diseases and certain mental disorders, which are considered to be inappropriate for childbearing, and requires physicians to give advice on marriage and reproduction. It also requires doctors to advise a couple to terminate a pregnancy if a hereditary disease is liable to result in a birth of a seriously sick or disabled baby. 40 The enactment of the law was regarded

40 The article concerned is Article 10. The official English version of it says: After premarital medical examination, physicians shall, in respect of the male or female who has been diagnosed with certain genetic disease of a serious nature
in the Western media as a kind of re-emergence of eugenics and attracted considerable criticism. It received a great deal of adverse publicity in the Western press. Article 19 of the law states that only under this legislation with the agreement of the pregnant woman or her guardian abortion can be carried out. Despite the inclusion of this article, some scientists considered that, because of China’s record in coercive birth control measures, it is hard to accept the assurance intended by the article (Pearson 1995). Chinese scientists claimed that the law was not motivated by racism but by a desire to reduce birth defects. This law embodies policy orientation and aims to prevent new births of inferior quality.

It has been considered that much of the confusion revolved around the Chinese word *yousheng*, which is a tricky word with dual meaning. In today’s Chinese language, *yousheng* routinely means ‘healthy birth’, and is always used with ‘youyu’, which means ‘rearing healthy children’. *Yousheng* can also be used to describe eugenic programmes that were practiced by the Nazis. ‘Eugenics’ also translates into Chinese as ‘*yousheng*’. In Chinese documents and the media it is more often used to refer to healthy birth. English translations of Chinese *yousheng* tend to reflect this latter meaning (Qiu 1999). The Chinese–English dictionaries give the explanation of Chinese word *you* as ‘excellent; superior [as opposed to inferior]’, and *yousheng* as ‘give birth to healthy babies’ (Hui 2004; Wu & Cheng 2005). It has same meaning as the Greek word ‘*eugenic*’ which means ‘well birth’ or ‘good in birth’. It is also consistent with Galton’s core eugenics doctrine of improving the stock of humankind by application of the science of human heredity. In this sense, the Chinese word *yousheng* could be translated to ‘eugenics’. But it is different from the ‘eugenics’ movement of the 1920s (Tang 2002; Wang 1999). The Chinese Encyclopedia of Medicine, which was edited in 1994, defines eugenics as: ‘a science for the improvement of human heredity, prevention of birth defects and raising the quality of the population by research applying genetic theories and approaches’ (Encyclopedia of Chinese 1994). Some geneticists consider that Chinese
yousheng is ‘eugenics’ in name only, and that, in practice, it is ‘prevention of birth defect’ and it does not go as far as would be expected of a eugenics movement (Ruan 2002). It was also claimed that there was not even one instance of a mentally ill person being forbidden to marry and have children despite extensive experience of Chinese psychiatric hospitals (Pearson 1995). There has been no research into the area of forbidding the marriage and reproduction of mentally retarded people. But, as of the date of a survey done on the condition of the mentally retarded population in the urban district of Nanjing in 2005, among 210 mentally retarded persons 71 reached legal marital age. Nine of these 71, one male and eight female, are married or have had a marriage history. It is quite difficult for mentally retarded individuals to become married, especially for males (Zhang 2006). Here, it is necessary to point out that Nanjing is a capital city, large and comparative wealthy. In rural areas, the situation could possibly be worse. In practice, after the coercive premarital medical check-ups were abolished in 2003, and the articles of Chinese Maternal and Infant Health Law on premarital medical examination are already virtually invalid.

‘Eugenics’ is still sensitive in China. As a national population policy, eugenics in China is not only a genetic term, also has certain social and political sense. A Chinese scientist has pointed out that, with regards to eugenics, one should be very careful not to equate the term eugenics with yousheng in Chinese, which means ‘give birth to a healthy baby’ (Chen et al. 1999).

GENETIC SERVICES AND PREVENTION OF BIRTH DEFECTS

An overview of the prevention of birth defects in China
Birth defects are a major cause of infant mortality in many parts of the world. At least 3.3 million children under 5 years of age die from serious genetic or partly genetic birth defects each year (WHO 2006). Most children who are born with major birth defects and survive infancy are affected physically, mentally or socially, and can be at increased risk for morbidity from various disorders. Birth defects have a substantial public health impact on mortality, morbidity, disability and healthcare costs (Rosano et al. 2000; Macleod 1993; Yoon et al. 1997). The birth of a child with congenital malformations may not only cause
suffering for the individual and the family, but also may increase the financial burden of an entire society. In China, according to the data of birth defects surveillance from the Ministry of Health (MOH), the rate of birth defects is about 11 per thousand. Every year, about 800 to 1200 thousands of children are born with congenital defects, which is about 4 to 5 per cent of the national total of births in China. The expense of treatment for Down’s syndrome yearly is about two hundred million Chinese Yuan which is about US$29.3 million. The MOH pointed out that birth defects and the disability resulting from birth defects have been major problems that affect population quality, and cause heavy financial burden for family and society (Luo 2005). In the Chinese context, under tension from a large population and the lack of adequate services for the care of affected infants, and under the population policy which aims to reduce quantity and improve quality of the population, the prevention of birth defect has become important. In 1989, the China Disabled Persons’ Federation (CDPF [zhongguo canlian]), in a letter suggesting the prompt adoption of measures to ensure healthy births and the prevention of the birth of handicapped infants, claimed that: ‘deformity brings about suffering to the handicapped, misery to the family, and a heavy burden to the country and society. In assisting the government and mobilizing the society, the CDPF devotes itself to the cause of handicapped, calls the whole society to act in the spirit of humanitarianism, and to understand, respect, care and help the handicapped, making them genuinely equal members in the society who enjoy rights of full participation. From the same principle of humanitarianism, CDPE insists that birth with deformity should be reduced. We call on the government and society to further strengthen the prevention of deformity, beginning with the prevention of congenital deformity and a reduction of the birth of unfortunate children. We maintain that the principle of humanitarianism is consistent with the improvement of the quality of our population’ (CDPF 1989). The Chinese government highlighted the prevention of birth defects, and the Ministry of Health and China Disabled Persons’ Federation collectively enacted the Action Plans of Improving Birth Quality and Reduce Birth Defect. The aim of this governmental plan is that the primary prevention measures coverage rate will reach at least 40 per cent by 2010, and that the high occurrence of birth defects will be basically controlled (MOH & CDPF 2002). In 2007, National Population and Family Planning Commission [guojia jishengwei] issued a document entitled Guidance of primary prevention of birth defects. This
document contains a flow diagram as detailed direction for how to address the primary prevention of birth defects, which highlights the role of genetic counselling. The flow diagram is as below:

Genetic counselling and primary prevention of birth defects

In 1947, genetic counselling was introduced to describe the relationship between clinical geneticists and those to whom they provide information about the aetiology, natural history and recurrence risks of hereditary disorders (Reed 1975). In 1975, the American Society of Human Genetics articulated its goal more broadly, defining genetic counselling as a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family (Ad Hoc Committee 1975). Accompanying the development of genetic counselling, there have been four models proposed: the eugenic model, the medical-preventive model, the decision-making model and the psychotherapeutic model (Baker et al. 1998). The eugenic model aims at counselling families with a history of ‘defective’ offspring not to reproduce. The medical-preventive model attempts to give families information and offer the option to avoid recurrence of a particular genetic disorder. Its aim is the optimization of population health, mostly through the prevention of birth defects. The decision-making model allows a more interactive type of counselling so that individuals are not only educated about their risks but also helped with decision-making on further testing and reproductive concerns. In the psychotherapeutic model, which became dominant during 1970s in Western countries, the genetic counsellor not only provides the individual with the necessary information but also helps the individual through any psychological and emotional difficulties. Genetic counselling itself cannot avoid the risk or recurrence of the genetic disorder and congenital malformation, but it plays an important role to enhance a couple’s awareness of prevention of births with a defect.

The MOH promulgated Guidelines for Genetic Counselling in 2003. The guidelines are official government rules, but they only regulate the genetic counselling related to prenatal diagnosis. The guidelines state that the main goal of genetic counselling is reduce birth defects. Clinical genetic counselling services are offered by clinical geneticists, such as obstetricians, gynaecologists or paediatricians with a genetic education background. According to the guidelines, the doctor/counsellor should give advice to the persons who are at high risk of giving birth to an affected child. Usually, such advice is concerned with the options of genetic tests and selective abortion. Although it is widely accepted in Europe that
genetic counselling does not aim to prevent couples from having children with genetic diseases and that the good clinical practice of counselling is more important (Harris 1998), in China, genetic counselling is treated as a general measure to prevent or reduce birth defects. In practice, besides the hospitals or healthcare providers with authorization from the Provincial Health Department to practise prenatal genetic diagnosis and to offer clinical genetic counselling, some local maternity and child care institutions [fuyou baojian yuan], also offer a kind of counselling services concerning genetics, productive health and good birth. These institutions are usually in the cities and the towns, such as the local maternity and healthcare stations [fuyou baojian zhan] and maternity hospitals. ‘Genetic counselling’ [yichuan zixun], in Chinese, does not only mean the clinical genetic counselling related to prenatal diagnosis and regulated by the governmental guidelines. It is widely used to mean all the counselling for prevention of birth defects and for healthy and good births. Sometimes it is also referred to as ‘eugenic counselling’ [yousheng zixun]. Actually, the counselling service offered by local Family Plan Commission and some low-level maternity and child care institutions is intended to generalize knowledge of heredity, offer healthcare in the perinatal period, and advise measures for primary prevention of birth defects before conception or at a very early stage of pregnancy, such as the intake of folic acid to reduce the risk of neural tube defects. From the report of a questionnaire survey on the 33 maternity and childcare stations in medium or small cities or counties, which was performed in 2002, 25 of 33 have counselling service (Ren & Wang 2002). Among these 25 institutions, 20 of them have a genetic counselling department or a similar department, but most of them have no practical guidelines or evaluation criteria. The counselling is offered by a premarital examination physician, obstetrician, paediatrician or healthcare doctor. In about one-third of the stations, the counselling physicians have not had any training in genetic counselling and do not or cannot offer a risk recurrence estimate. For the cases that cannot be dealt with in primary institutions, such as those counselees who have a family history of genetic disorder or who already have an affected child, the doctors will advise them to visit a genetic counselling clinic in a high-level hospital in a large city. In fact, the availability of counselling service in 41 In my paper The Practice of Genetic Counselling—a Comparing Approach to Understand Genetic Counselling in China, I describe the situation of genetic counselling offered by authorized hospitals or healthcare providers.
medium-sized or small cities is not widely known. Many pregnant women think it is not necessary to resort to genetic counselling if there is no family history of a genetic disorder. Some have never heard of genetic counselling. As for recessive heredity disorders, such as thalassaemia in the high-incidence area in south of China, many carriers do not realize they are carriers till they have an affected child.

**Genetic screening, testing and birth defect intervention**
The cost of caring for a severely disabled child is high, and the fate of the child after the parents are no longer able to provide care is a source of great anxiety, even in systems with national health insurance (Kerr & Shakespeare 2002). In China, there is no national medical health coverage system [guojia gongfei yiliao tixi]. The existent healthcare insurance system is under developed and usually the inherited diseases are excluded. In fact, the majority of the population pays medical costs themselves, except for government officials and employees in state-owned units. Illness has a close correlation with poverty, especially in rural areas in China. It has been shown that the costs of medical services are a major factor influencing hospital utilization by the poor, and also that medical costs contribute to families falling into poverty (Kaufman & Fang 2002). For the absolute majority of Chinese families, the financial burden of medical treatment for a child born with a serious genetic disorder is so oppressive that it can hardly be afforded, such as with thalassaemia, which requires lifelong treatment. It requires considerable time and energy to care for an affected child. Sometime one of parent has to stop working, and the costs of caring becomes a considerable economic burden. Additionally, deficient public welfare and the potential social stigma for the families with inherited diseases make prospective parents worry and suffer more. To some extent, having a child with serious hereditary handicap is miserable for a family. The former minister of Public Health, Chen Minzhang, pointed out that ‘the cost of looking after those with hereditary handicaps was enormous, imposing a heavy burden both on the state and on millions of families’ (Wang 1999). Under the one child per couple population policy, Chinese parents have more expectations for their offspring. Having a healthy baby is the minimum wish for prospective parents.

The National Population and Family Planning Commission launched a national project
entitled *Project of Intervention with Birth Defects* in 2001. The project aims to place increased stress upon the prevention of and intervention in six genetic disorders, such as neural tube defects, congenital rubella syndrome, Down’s syndrome, Duchenne muscular dystrophy (DMD) and thalassaemia. All the selected diseases have a high occurrence in various parts of China, and technically can have effective prevention measures. The project encourages the application of primary screening, prenatal diagnosis and subsequent selective abortion to prevent birth defects. This is treated as a major project that benefits both nation and individual [liguo limin]. Based on the respective actual situations, the local governments and health authorities have feasibility plans and the local hospitals and healthcare institutions identify the specific measures to prevent and reduce birth defects. Recently, Shanghai started a plan offering free prenatal screening on Down’s syndrome for ten thousand pregnant women. The Shanghai municipal government hoped that this plan might help to reduce birth defects due to Down’s syndrome (Qiu & Xu 2009). In practice, premarital screening, which can be combined with the carrier test, is often selected as the preferred approach in the high-occurrence area of a certain genetic disorder, because it allows carriers the widest range of choice, including using information of carrier status when choosing a partner. In the thalassaemia high-occurrence area, some cities such as Nanning and Guangzhou had listed thalassaemia carrier screening as a necessary item for premarital check-ups. After the lift of compulsory premarital check-ups according to the new *Rules for Marriage Registration* in 2003, the number of couples taking premarital check-ups has dropped. Some local governments have taken measures to encourage people to take a premarital check-up. For example, the government of Guangzhou has offered free premarital check-ups, and the Nanning government, in 2006, launched a project of free carrier testing for thalassaemia for a hundred thousand newly married couples in the rural area (Li 2006).

In practice, the licensed hospitals also develop prenatal screening and use prenatal diagnosis to reduce birth defects. Gradually, public awareness of the prevention of birth defects has been enhanced, and the number of people attended prenatal screening is increasing. For example, the Provincial Maternity and Child Care Hospitals of Hubei province declared their 12-year statistical data of prenatal screening. From 1997 to 2008,
these hospitals conducted 167,171 screening cases. Among them 4,937 congenital defect cases were diagnosed, and the rate of the birth defects was 2.9 per cent. However, during the year of 2008, these hospitals conducted 15,047 screening cases, diagnosed 1,294 congenital defect cases, with a rate of birth defects of 8.6 per cent (Fang & Wen 2009). The number of cases screened and the rate of birth defects in 2008 were doubled compared to 2007. One clinical geneticist in these hospitals considered that such changes show that the prevention consciousness of pregnant women had increased and the diagnosis technology had been improved. Another example is the intervention in birth defects with thalassaemia in Guangdong. Guangdong province is a high-prevalence area for thalassaemia, where the thalassaemia gene frequency is 2.5 per cent. More than 160 neonates with thalassaemia major would be expected to be born each year, considering an annual birth rate of one million in this province (Xu et al. 2004). Qualified large hospitals that can offer prenatal genetic testing for thalassaemia play an important role in the reduction of births of children with thalassaemia. One such example is the prenatal screening programme for thalassaemia in the Prenatal Diagnostic Centre of Guangzhou Maternal and Neonatal Hospital. Since 1993, this centre has conducted a hospital-based prenatal screening programme for thalassaemia, which had an objective to control the birth of thalassaemia children in Southern China. Pregnant women are screened for a thalassaemia trait at their first presentation for prenatal care at the hospital, unless these investigations have already been performed elsewhere. The partners of women with a positive screening test are also required to take a screening testing. A pregnancy with both partners being carriers of the same type of thalassaemia is considered a risk, and genetic counselling services and prenatal diagnoses are offered. During the period January 1993 to May 2004, 545 fetuses of 540 mothers underwent prenatal genetic testing at this centre. These 540 pregnancies were at high risk for producing a child with thalassaemia or a thalassaemia carrier. The tests results shows that 154 were found to be normal, 257 were carrier, and 134 were affected. In total, 133 pregnancies with affected fetuses, except for one twin pregnancy with one affected and one unaffected fetus, were voluntarily terminated (Li et al. 2006). The clinical geneticists in this centre consider that the prenatal diagnosis developed in this centre has proved to be highly effective during the 11 years from 1993 to 2004. The centre managed to prevent 133 cases of birth defect with severe thalassaemia, which would
have led to expensive treatment if pregnancies had continued, and lead to a marked reduction of birth defect of thalassaemia in that region.

Additionally, there is an example regarding the intervention in birth defects with Down’s syndrome by prenatal genetic screening in Kunming, Yunnan province. The natural incidence, without intervention, of Down’s syndrome is about 1 in 700 in the area (Cuekle 1995; Binkert et al. 2002). The Genetic Diagnosis Centre of Yunnan No.1 People’s Hospital has performed a study on the incidence of Down’s syndrome and the intervention by prenatal screening. The study result shows that from 2000 to 2002 in this hospital the number of outpatient pregnant women was 7761. Among them, 1886 accepted prenatal screening for Down’s syndrome. The prenatal screening rate was 24.3 per cent (1886 of 7761). In the same period, 7063 live infants were delivered, and among them there were seven cases of Down’s syndrome. Total incidence of Down’s syndrome in this period was about 1 in 883. During 2003 and 2004, 81.4 per cent (4725 of 5790) outpatient women in the first and second trimesters of pregnancy were accepted for the prenatal screening and three cases of Down’s syndrome among 5273 live births were delivered. The incidence of Down’s syndrome decreased to about 1 per 1,758. The geneticists who conducted this study view prenatal screening is an effective method to discover the high-risk cases during the first and second trimesters, and they believe the study shows that increasing the screening rate in the first and second trimesters of pregnancy could reduce the incidence of Down’s syndrome in live births (Zhu et al. 2005).

DISCUSSION

With the aim of improving population quality, primary prevention measures and genetic services play very important roles in preventing and reducing birth defects. However, there are some issues involved in this field that are worth considering.

Access to and public awareness of primary prevention

The economic development between different areas and between town and country is
imbalanced in China. The allocation of medical resources is also imbalanced. Genetic services and prevention measures are much better developed in cities than in rural areas. In some large cities, usually the local maternity and child care stations have a special course of ‘well birth and well bear’ [yousheng youyu ketang] for married couples who plan to have a child or are already in early pregnancy. Usually, the course takes one day for a charge of 40–50 Chinese Yuan (about US$6–7) (Shao 2007). Under the Chinese supervision of family planning and maternity and child care system, married couples should apply for a reproductive permission certificate [zhunsheng zheng, which authorizes a planned birth] before pregnancy or at an early stage of pregnancy. Taking such a yousheng course is required for application for reproductive permission. Through such a course, the prospective parent can get access to genetic knowledge and to common counselling for yousheng.

As mentioned above, the coverage rate of primary prevention measures for birth defects is still low now and is expected to reach 40 per cent in 2010. Even if this rate is achieved, there still will be about 60 per cent by area under the primary prevention measures, and possibly most will be in the rural areas. In fact, however, births with congenital malformation and births with genetic disorders are relatively more common among the Revolutionary Old Base generation [geming laoqu]. The Chinese revolution originated in remote mountainous areas, which were called Revolutionary Old Bases, the frontier, and countryside in China (Dickson 1994). Because of comparative underdevelopment and geographical isolation, such areas are usually economically disadvantaged areas, and medical resources are comparatively deficient. In some economically poor places, healthcare needs are unmet, and, commonly, the public is largely uninformed about genetic risks and the possibilities of prevention. In one area, the Dabie Mountains (one of the Revolutionary Old Bases), inbreeding was estimated to be the source of approximately 37.5 per cent of severe mental retardation (Qiu 2004). According to the official sampling investigation data of the China Disabled Persons’ Federation, the average incidence of mental retardation in China is about 12.7 per cent. The incidence in cities is 6.6 per cent, in towns 11.1 per cent, and in the countryside 14.1 per cent (CDPF 2008). The incidence in the country is thus higher than in the city. People in comparatively remote areas have little knowledge of inheritance, and do not know the hereditary feature of a recessive inheritance. In practice, many people simply take for granted
that ‘inherited disease’ is transmitted to offspring by the parents who have ‘inherited disease’. For recessive genetic disorders, many people even in the high-occurrence areas have no idea about carriers of genetic disorders and the inherited risk of a person who is a carrier. One couple, who live in a village of Bobai in the county of Guangxi province and have a child affected by thalassaemia, explained their opinion:

   We never heard about thalassaemia before my son’s ‘anaemia’ symptom was diagnosed. We think that both of us are healthy and have no disease; even the relatives of our family have no disease. It is natural we should give birth to a healthy baby. Nobody told us and, you know, we have less schooling [mei duoshao wenhua]. We try to earn money to have a baby, to raise a family. We do not know many things that are far away from our life. If we know we are carriers, if we know we are at such risk, we think we will pay more attention. We think we will take a test, despite its expense.

The mother of a boy with DMD, who comes from Suining county of Anhui province, told of her experience:

   After my son was diagnosed with DMD, I understood a few of the inherited characteristics of DMD. I told my female cousins that possibly they are at risk. But they do not take it seriously, and even do not believe me. They thought they are fine and without any disease, and there is no reason to give such a birth. They thought what happen to me was because of my bad fate [ming bu hao], and they believe that they may not have such bad luck.

The National Population and Family Planning Commission [guojia jishengwei] lists some feasible methods of public education in basic genetics knowledge, such as the Internet, TV programmes, newspapers, leaflets, blackboard newspapers and other media. One geneticist working in the genetic counselling clinic of a famous hospital said she met a girl who applied for carrier testing for DMD in her clinic. The girl came from a village but worked in a city.
She had a cousin, the son of her mother’s sister, who had been suffering a walking problem like the symptom of DMD for years. The geneticist said she was very gratified the girl can find information about DMD on the Internet and resort to clinical genetic counselling before she gets married. However, the situation is that most of rural areas have no access to the Internet and, moreover, most of the people there are unable to use a computer.

The popularization of genetics, which requires large amounts of human and material resources to support it, is difficult to implement in the comparatively poor areas of China. Additionally, although the population policy also calls for ‘well bear, well rear’[yousheng youyu], the actual work of family planning at the local level pays much more attention to reducing the birth rate than to the primary prevention of birth defects, specially in the villages, which is the lowest official administrative unit of rural areas. In fact, there is a well-organized official network to administer population policy in rural China. The National Family Planning Commission implements both vertical control of policy from the top and horizontal integration of services at the local level, which has been described as follows:

At the local level, the family planning cadres maintain intimate contact with village officials and continuously monitor the local birth planning official, usually the Women’s Leader, of the Village Council. The Women’s Leader is a full-time cadre who spends approximately half of her time doing a mixture of family planning work, which includes recording the number of married women by parity, and calculating how many couples are allowed to have first children and second children. She is expected to know which women are using contraceptives, what methods they use, and which women have become pregnant (Davin 1985).

In practice, local level of the Family Planning Commission and the family planning cadres focus more on the control of approved birth and expend most of their effort to stop or reduce unapproved births in their prefecture. Many persons who take charge of family planning in small villages do not understand well the inherited features of the serious genetic disorders listed in the Project of Intervention with Birth Defects. Most of the propagandizing regarding population policy is about the advantage of having one child and the disadvantages, usually
the economic burden of having more children. Easily available information on basic genetics and the prevention of birth defects is still uncommon in the countryside in China. One of my interviewees, a well-known geneticist in China, insisted that the genetic measures for diagnosis and prevention of genetic disorder should be accessible to everyone, regardless of the ability to pay, and especially for those most urgently in need.

The individual’s right not to know personal genetic information

Carrier testing is a method used to identify individuals who carry a genetic abnormality that does not affect the health of the individual in question but which increases the risk of producing offspring with a serious genetic disorder, such as thalassaemia or DMD. Although carriers are asymptomatic and do not develop any signs of the disease, they pass the genetic disorder to their offspring. Information regarding being a carrier may be useful in making reproductive plans. Thalassaemia and DMD are considered preventable if the carrier status can be confirmed, and pregnant women are tested for these conditions, followed by selective termination of pregnancy if the fetus is affected. Thus, premarital carrier testing is treated as the first line of defence for primary prevention of birth defects.

In order to reduce births affected by genetic disorder, carrier testing, especially prenatal carrier testing, is strongly encouraged. However, an important issue should not be ignored—the right of the individual not to know personal genetic information. Usually, genetic counselling is recommended before and after the test to prevent confusion over the difference between being an asymptomatic carrier and being affected by a disease. Although genetic counselling aims to minimize adverse psychological reactions, it does not prevent carrier screening from creating opportunities for racial discrimination, as happened with sickle cell screening in the USA in the 1970s (Prainsack & Siegal 2006). Identified carriers can harbour continuing regrets and worries. In the Chinese context, in which there is often no professional genetic counsellor and the genetic counselling is more like the medical-preventive model, the situation might be worse for identified carriers. The information of individuals with carrier status benefits the prevention and intervention related to birth defects, but at the same time adverse impacts might be caused by the disclosure of carrier status. Take the premarital thalassaemia test as an example. When couples come for a test, the best expectation for them
both is to be thalassaemia free, but they should also be prepared to face the various possible ‘bad’ or ‘not best’ results (e.g. either partner, or both, is identified as a carrier) and to consider the problems and decision-making related to each result. In fact, to the couple, each possible result may create a psychological burden and confusion, which may disturb the prospective bride or groom, and possibly also the parents of the couple. If no proper psychological support is available, such adverse outcomes may interfere with the prospective marriage and hurt both of the couple and even their parents. Additionally, the discrimination caused by the disclosure of carrier status may make it difficult for the indentified carrier to find a partner. Considering the adverse impact on the individuals, the right to know or not to know personal genetic information should also be respected, even though carrier testing is officially encouraged as a measure for the prevention of birth defects.

**Reproductive pressure, responsibility and choice**

In many parts of China, giving birth to children and carrying the family line for the husband’s family is treated as a woman’s duty. Since family planning has become a policy of national priority, especially since 1980 when the one-couple one-child policy began in China, the desire for healthy children has increased. Women are expected not just to give birth to a baby, but to give birth to a healthy and intelligent baby. Additionally, the national ‘well bear, well rear’ *yousheng* policy, with the aim of improving population quality, has raised ‘good’ births to a high level as the factor that determines the Chinese national quality [*zhonghua minzh de sushi*]. Healthy and intelligent babies are not only the expectation of individual and family but also the expectation of society and the nation. Additionally, China is a socialist country in which the mainstream ideology regards each individual as a component of the whole society or the nation, and advocates that individuals’ interests should properly be subordinated to the interests of the whole society or nation (Qiu 1993). In 1981, the People’s Health Press published a booklet entitled *New Knowledge of Eugenics* [*you sheng xin zhi*]. The booklet is the first book in mainland China that systematically relates the history of eugenic knowledge since 1949, the time the PRC was established. The first sentence of this booklet is: ‘*parent, family, society and nation all hope the next generation healthy, intelligent and beautiful*’. Later, this sentence was used as a propaganda slogan on huge billboards in Beijing and others
cities (Ruan 2002).

Greater expectation adds much to the reproductive pressures placed on women. Additionally, the women are considered to be responsible for ‘bad births’ or ‘inferior births’, and ‘failure’ in the light of these high expectations may also cause self-condemnation and guilt.

As the main measures for the prevention of birth defect, genetic services such as prenatal screening and prenatal testing have been developed in some qualified hospitals. As more genetic disorders become identifiable by genetic testing, and testing becomes more available in the clinical setting, participating in genetic screening and undertaking genetic testing are increasingly seen as what responsible and good parents should do. Usually, pregnant women would like to take tests as long as they will prevent birth defects and the doctor advises or recommends them to be taken. Even though the cost is high, women think that the money spent on prevention is worthwhile. Genetic testing followed by selective abortion has already been considered to be an effective way to reduce the occurrence of birth defects. It is reported by the Chinese Yousheng Association [zhongguo yousheng xiehui] that 248 pregnancies with defects, most of which are congenital heart disease, spina bifida and cleft lip and/or palate, were diagnosed through prenatal screening and prenatal testing in Beijing in 2006, and 248 occurrences of birth defects were prevented by termination of the pregnancies. The association pointed out that this achievement was estimated to reduce approximately 59.6 million Yuan (about US$8.52 million) of financial burden (Huang 2007). The Chinese Yousheng Association praised the fact that prenatal diagnosis lightened the burden on the families and state, and created a harmonious life for society. In fact, pregnant women are pressured by economic circumstances and social expectations. The high expectations of family and society, reproductive pressures and the responsibility for give a good birth, and the foreseeable financial burden that may be caused by a baby with a birth defect, limits their choices and decisions during the receipt of genetic services. Actually, in certain social contexts, their individual choices also reflect social expectations, and tend to correspond with the expectations of society.
Yousheng ideas and genetic discrimination

Under social circumstances in which ‘good birth’ and ‘population quality’ are excessively emphasized, it is inevitable that such stress sharpens the problems of social stigma and discrimination. The opposite of good birth is ‘bad birth’ or ‘inferior birth’ [lie sheng]. The more ‘good birth’ is encouraged, the more ‘bad birth’ or ‘inferior birth’ is discouraged, and, at the same time, the limits of so-called ‘bad’ or ‘inferior birth’ become more extended. Thus, as mentioned, the Chinese Yousheng Association reported and praised prenatal diagnosis for reducing the occurrences of birth defects, including those of cleft lip and palate. In practice, persons suffering from a severe illness, and their close family, are often socially stigmatized (Sartorius 1997). In fact, it is very difficult for the disabled to lead a ‘normal’ life, and it is challenging for the disabled to get married, as the disability sets them apart from their friends and neighbours (Kohrman 1999). Excessive expression of yousheng ideas implies that what is not related to ‘good birth’ is a burden on the family and state and is unwelcome in Chinese society, and this may increase the stigma and discrimination against people with genetic disorders.

As a national policy, yousheng can be understood as the hope to reduce defective births, and it is quite different from Nazi Germany’s eugenics, which is essentially racism. However, during its implementation, some local enforcement regulations take an approach similar to eugenics. One such approach is to focus on reducing the number of individuals within the population who suffer from physical deformities or mental retardation by limiting marriage and reproduction by compulsory means. An example of the regulatory approach is the regulation of Gansu province, called the Regulation on Prohibiting Reproduction of the Dull-witted, Idiots or Blockheads [chi, dai, sha ren], which was promulgated by the Fifth Session of the Standing Committee of People’s Congress of Gansu Province on 23 November 1988 (Lei et al. 1991). According to this regulation, the definition of ‘dull-witted, idiots or blockheads [chi, dai, shan ren]’ is: congenitally caused by familial inheritance, inbreeding or parents under external influence; mental retardation at middle or severe degree with IQ below 49; behavioural disorders in language, memory, orientation, thinking, etc. It prohibits the reproduction of the dull-witted, idiots or blockheads. The dull-witted, idiots or blockheads
may be married only after sterilization. If both partners are dull-witted, idiots or blockheads, only one has to be sterilized; if only one of the couple is dull-witted, idiot or blockhead, only he or she may be sterilized. Another example is the regulation of Shaanxi province called Measure on Limiting Reproduction of the Dull-witted, Idiots or Blockheads. This regulation was promulgated on 31 December 1991 and gave the similar definition of dull-witted, idiots or blockheads and similarly limited their marriage and reproduction. At the time of promulgation of this regulation, Shaanxi provincial government enacted a government document to enforce the regulation, in which it pointed out that limiting reproduction of the dull-witted, idiots or blockheads is a very important measure to actualize yousheng youyu and to prevent the reduction of population quality. Additionally, some opinion labelled physical deformity and mental retardation as ‘inferior birth’, and regarded parents with severe genetic diseases, severe schizophrenia and inbreeding as ‘parents without reproductive value’ (He & Shi 1989; Wu & Wen 1990; Zhang 1990; Chen et al 1992). These terms are prejudicial with regard to handicapped persons. To a larger extent, these regulations and opinions are eugenic in nature, although they are different from racial eugenics. These two regulations had been annulled retrospectively after being valid for 14 years and 17 years. Gansu’s regulation was annulled on 22 January 2002 and Shaanxi’s regulation was annulled on 2 September 2008. In fact, although such regulations have been annulled, their influence and implications can not be changed in a short time, and they continue to aggravate discrimination against the mentally retarded and the handicapped.

CONCLUSION

In consideration of the serious situation regarding birth defects in China, the government has implemented an intervention project to prevent birth defects and improve of the quality of newborn babies, which is the purpose of eugenics in China. Some of the advanced genetic

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42 The full text of this regulation can be found online: [http://www.pjssj.com/Article_Show.asp?ArticleID=2](http://www.pjssj.com/Article_Show.asp?ArticleID=2). [Accessed date March 2010].

43 This government document can be found online: [http://www.51zy.cn/44717583.html](http://www.51zy.cn/44717583.html). [Accessed date March 2010].

and reproductive technologies have been used in clinical genetic services in some qualified hospitals. In practice, the genetic services such as premarital, pregestation and prenatal genetic screening, counselling and testing play important roles in the prevention of birth defects, and in some places the prevention programmes are regarded as successful through the use of selective abortion of pregnancies with identified defects. Prevention of birth defects has developed rapidly. Avoiding the birth of a baby with a birth defect benefits families and individuals by helping them to have an easier life, and also reduces the associated financial burden. However, in the national rush to reduce birth defects, some individual rights and choices involving personal genetic information and decision-making are being ignored. An insufficiency of psychological support and humanistic care in genetic services still exists widely in China. Overmuch stressing of the financial burden placed on the family and society by people with birth defects has increased reproduction-related pressures on women and may increase genetic discrimination.

Actually, there are some concerns related to birth defects and population quality which require rethinking. For example, in a social environment in which there is a high expectation for the production of ‘superior’ offspring and the prevention and reduction ‘inferior birth’, an appropriate definition and understanding of a ‘normal birth’ deserves careful consideration. Additionally, the term ‘population quality’ needs clearer definition. For example, what is the meaning of ‘quality’ with regard to a so-called ‘normal’ person, or to a person who is affected by, or is a carrier of, a genetic disorder or congenital defect. And what is the meaning of ‘quality’ for the nation and society? Such questions deserve serious thought.

Finally, I would like to quote a Chinese scientist, one of the founders of Chinese eugenic science:

Science cannot give a model of a good gene. Life is varied and the charm of life exists in its diversity. People with a ‘defect’ are also part of the diversity. It is impossible to create perfect population, and it is unnecessary (Ruan 2002).