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

Sui, S.

Citation for published version (APA):
CHAPTER SIX

CASE STUDY: GENETIC TESTING FOR DUCHENNE MUSCULAR DYSTROPHY AND THE VULNERABILITY OF FAMILIES WITH DUCHENNE MUSCULAR DYSTROPHY

This chapter concerns the consequences of the application of genetic counselling and testing for Duchenne muscular dystrophy (DMD), which is a sex-linked genetic disorder. The chapter aims to provide a better understanding of the economic difficulties, psychological distress, self-contempt and family stigma that families are confronted with in Chinese contexts. In present-day China, DMD-affected families are not only undergoing economic hardships but also face heavy psychological pressure as a result of family stigma. Meanwhile, Chinese values attached to healthy offspring and traditional boy-preference increase the psychological burden, especially of the mothers of children with DMD. Families affected by DMD are vulnerable, and the existing social discrimination aggravates their vulnerability. This chapter shows that there is room for social welfare and support to play a greater role in reducing the vulnerability of these families.
INTRODUCTION

In China about 650,000 affected boys suffer from Duchenne muscular dystrophy\(^{31}\) (CRCF 2005). Duchenne muscular dystrophy (DMD), the most common childhood muscular dystrophy, is a lethal X-linked genetic disorder, which affects approximately 1 in 4500 live male births (Garner-Medwin & Sharples 1989). DMD carriers pass it on to one-half of their sons, which means 50 per cent of male offspring will be affected, and to one-half of their daughters who become carriers (Harper 2004:100). This chapter concerns the consequences of the application of genetic testing and genetic counselling for this sex-linked genetic disorder. Genetic counselling and testing for DMD has important implications for reproductive decisions and life planning decisions, which are also affected in China by economic conditions, access to the healthcare system, the Chinese family-planning policy, Chinese reproductive views and Chinese culture. This case study of DMD aims to acquire a better understanding of economic difficulties, psychological distress, self-contempt and family stigma that families are confronting with in Chinese contexts. Based on the complexity of conditions of families with DMD, this study rethinks some related social issues such as genetic discrimination, the distribution of health resources, the reliability of medical information, the access to basic knowledge and of medical treatment of prevailing genetic disorders, the impact of genetic technology on reproductive decision-making, social support and help for vulnerable groups.

This chapter is based on the research results of an empirical survey conducted from September 2007 to February 2008 in China. During the survey, basic information about genetic testing for DMD was collected in China through archival study and Internet research. The original data are obtained from interviews and participant observation. During fieldwork

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\(^{31}\) Duchenne muscular dystrophy is a recessive disease. The female is the carrier and transmits the disease genetically to offspring, but does not develop it. DMD is caused by the lack of dystrophin, which is a protein found in the cell membrane of muscles. Boys are normal at birth and only begin to show signs of the muscle wasting disease when they are 3 or 4 years old. They experience increasing difficulty walking because of progressive weakness, with loss of ambulation, and are often wheelchair bound by 11–12 years old. The muscle deterioration is continuous and they usually die in their late teens, or early twenties, because no effective treatment is yet available (Parsons et al. 2000).
the author interviewed five geneticists and six doctors working in genetic counselling clinic as genetic counsellors, and sat in at two sessions of genetic counselling in both the paediatric department and maternity department of a hospital in Beijing. The sessions target genetic disorders in general, not just DMD, but one session usually includes two or three consultations with families with DMD from all of the country. As a ‘noviciate doctor’ (jianxi yisheng),³² I collected first-hand information of the practice of genetic counselling for DMD. Such experience provided access to boys with DMD and their families. The method of deep interview was used with ten families with DMD, and four families with DMD were selected as case studies, with which regular contact was maintained. The research method of participant observation yielded a better understanding of the daily routine and the problems that families with DMD experience in daily life.

THE PRACTICE OF GENETIC COUNSELLING AND TESTING FOR DUCHENNE MUSCULAR DYSTROPHY IN CHINA

Although there is no cure or effective treatment available for DMD, an early diagnosis is ideally made, primarily to offer genetic counselling, and, where possible, prenatal diagnosis. When diagnosis of DMD takes place, the family is warned that DMD is an inherited disorder and that the sisters and cousins of the affected boy have a high risk of being carriers. Improving the identification of carriers is aimed at reducing the number of boys with DMD being born in affected families, as is the case in Europe (Emery 1991). In China, it is the genetic counselling clinic, usually in the paediatric department or/and maternity department of the hospital, that offers genetic testing for diagnosis. For the prenatal genetic counselling and testing, it is usually in the genetic clinic of the maternity department because the test sample and the amniotic fluid are collected by amniocentesis, an operation usually performed by a maternity doctor in the maternity clinic. Guidelines for Genetic Counselling promulgated by the Chinese Ministry of Health (MOH) in 2003 regulate the basic requests,

³² Noviciate doctors in China usually are medical students who are in the last year of university education. Clinical experience of noviciate doctors is compulsory before graduation. Thanks to the introduction of one respected professor and consent from the doctors and patients, I had the chance to carry out my survey as a ‘noviciate doctor’ in the clinic.
principles and procedures of genetic counselling in China. According to these guidelines, genetic counselling should be offered by clinicians who have a genetics knowledge background. In practice, the counsellors in the genetic counselling clinic usually are clinicians such as paediatricians and obstetricians with a background in genetics. More details about the qualifications for counsellor could be found in the section ‘The current practice of genetic counselling in China’ of Chapter Three in this thesis. In China, genetic counselling is combined with testing, and it is the doctor who works as a counsellor, prescribing the testing for clients/patients. Because of the advanced nature of the technology of genetic testing for DMD and the qualification should be authorized by the Provincial Health Department.33 There are only some hospitals that are well known for their genetics departments and are authorized to offer DMD genetic testing, such as Beijing Xiehe Hospital, Hunan Xiangya Hospital and Shenyang Medical University No.2 Hospital. Most of the families that come to the counselling clinic obtain information on access to genetic counselling/diagnosis from doctors in their local hospitals.

In the genetic counselling clinic, collecting genetic information is the first important step. In counselling practice in the UK, general strategy demands that at least basic details of both sides of the family are taken, even in a dominantly inherited disorder clearly originating from one side. Taking details about both sides may help to avoid feelings of guilt or blame resting exclusively on one member of a couple. This may always be an important factor, but it is particularly important in some cultures and social situations (Harper 2004:7–8). This view is also accepted by Chinese genetic counsellors and is applied to genetic counselling, in particular for X-linked inherited disorders such as DMD. In the counselling clinic for DMD, doctors make efforts to avoid spreading feelings of guilt and blame among husband and wife. Usually during the first consultation at the counselling clinic, symptoms of the boy who is under suspicion of DMD are checked, the disease history of both partners’ families is taken, and the doctor provides an initial diagnosis. This initial step is complemented with genetic testing for confirmation. During the consultation, the doctor will explain some basic

33 According to The Measures for the administration of prenatal diagnosis technology, which is a document promulgated by the MOH in 2003, only hospitals and healthcare providers that are qualified by Provincial Health Department can practice prenatal genetic diagnosis.
knowledge of DMD in simple terms, though the doctor usually does not mention the X-linked inherited character to the couples directly. Usually, in practice, the doctor emphasizes that spontaneous mutations also can result in the birth of boys affected with DMD, avoiding the issues of ‘responsibility’ for the boy’s affliction. During counselling sessions for DMD, the doctor working as genetic counsellor in the clinic also takes into account the position of women and the stability of the couple’s marriage and their family. The survey among geneticists also showed that genetic counselling for X-link disorders should try to play down [danhua, a literal translation to English is ‘de-salt’] the issue of ‘individual responsibility’ for the genetic disorder.

Doctor S, a geneticist cum doctor working in the genetic counselling clinic, exemplifies genetic counsellors in China:

In the counselling of a family with DMD, I usually do not choose to directly tell the couple, especially the husband, the inherited character of DMD. Before, there have already been some examples where after the husbands know their boy’s disease is because of the wife’s gene, blame the wife very much and divorce. Usually the wife feels guilty and self-condemned in such a condition; if the husband requires divorce, she has no choice. You know, it is very miserable for a single mother with an affected child. Considering these, I think sometimes a ‘fog bomb’ is better (yanwu dan, meaning something is not clear or to make the clear thing hazy), especially for the families that come from the rural areas, where the Chinese traditional ‘boy-preference’ reproductive opinion is prevalent, compared with urban places. If the family does not ask, I don’t tell them, but if they ask I give them a ‘fog bomb’, such as that they can both be the ‘cause’ or that there could have been a mutation. In fact, there is a possibility of genetic mutation, and it is difficult to give a definite answer. I hope the couple have more psychological preparation, and understand that the most important thing is to face the fact instead of guiltiness or blaming. [Transl. S. Sui]
the prescription of genetic testing, and the blood sample will be collected from the affected boy and both parents. The doctor suggests that the couple come to the genetic counselling clinic for a second consultation, usually two months later, and at that time the genetic testing result is ready. For the second consultation, if the resulting diagnosis is DMD, the doctor will give directions on how to take care of a boy with DMD. Stretching, massage and low-impact sports are usually recommended by doctor to the parent of such a boy. According to the family pedigree, the doctor gives advice that the female members of the family, who have a high risk of being carriers, ought to take the test. The doctor also advises the couple to make sure prenatal genetic testing is available before they plan a next pregnancy and take a prenatal genetic test during the next pregnancy.

CASES SELECTED FOR UNDERSTANDING THE COMPLEX CONDITION OF FAMILIES WITH DUCHENNE MUSCULAR DYSTROPHY

This study is based upon conversations with approximately thirty DMD consultancy clients, including affected boys, carrier women, and their families. From these, three cases were chosen as case studies and one, based on Internet research, was added. The four cases were selected to offer insight into the variety of the circumstances in which families of boys with DMD have to make decisions, the complexity of conditions in which such families live their daily lives, and the vulnerability of these families in China. The four case studies are analysed in the following sections.

Case 1: Ms S wants a genetic test

This case concerns the decision-making regarding the termination of pregnancy and the expectations of this family about genetic technology. The case shows that family history did not get the attention deserved in this family.

Mrs S, a woman seven month’s pregnant from Qingyang, a small city in Gansu province, applied for prenatal genetic testing for DMD. Her application was rejected because it was too late to take amniocentesis at seven months’ gestation, which is very dangerous for both mother and fetus. In practice, amniocentesis is commonly used as a measure to collect
samples for prenatal genetic testing. Amniocentesis is performed at 10 to 20 weeks’ gestation in current clinical application in China. Mrs S and her husband made the decision to take a carrier test and decided to terminate the pregnancy if the test indicated her to be a carrier of DMD. Although one of her old brothers, two cousins and one nephew died as a result of DMD, she is not aware of her family history regarding DMD. She ascribes her little knowledge about DMD in her family history to the large size of her family and to having almost no memory of her deceased elder brother. When the son of one of her cousin, a four-year-old boy, started to show the symptoms of DMD and was diagnosed with DMD after taking a genetic test, she panicked and went to the genetic counselling clinic to ask for a test. The boy, from Quzi town, Huan County in Gansu province, is the only affected live male in this big family. His father and mother work as ‘farmer workers’ in Guangzhou. ‘Farmer workers’ [nongmin gong] are farmers who have temporary jobs in the city. His parent left him with his grandparent. Now, Mr S, an old brother of Mrs S, who is a surgeon in a hospital in Beijing, hopes that the genetic testing can help to eliminate this disorder from his big family by preventing the birth of affected boys and new carriers. (Interview with Ms S, her husband and her brother on 16 and 20 November 2007)

**Case 2: Ms L and her husband want to have a daughter**

This case illustrates ‘gender preference’ in reproduction, the social isolation of boys with DMD, the psychological pressure and the difficulties the families of such boys in China.

Mrs L is from Gaozuo Town, Suining County, in Jiangsu Province. Mrs L is the mother of a 9-year-old boy with DMD. Two years ago, this family took a genetic test in a hospital in Beijing, and the boy was diagnosed with DMD. Now the boy can no longer walk well. His situation gets worse when he wears thick clothes in the cold winter. Every day his father or mother carry him to school and get him home using a manual tricycle [renli sanlun che]. On this occasion, Mrs L came to the genetic counselling clinic when she was twelve weeks pregnant. She and her husband fervently hoped for a girl and decided to take a prenatal genetic test in Beijing. One month after the test sample was collected through
amniocentesis, they received the test result that showed the fetus to be female. The couple was very happy when they heard the result and they did not care greatly whether their future daughter was a carrier or not. Mrs L and her husband know about the inherited character of DMD. Mrs L has told her sisters and cousins about the inheritability of DMD and the possibility of their being a carrier, but they do not believe her and do not take it seriously. (Interviews with Mrs L and her husband on 27 November 2007 in a hospital in Beijing, on 6 December 2007 when Mrs L underwent amniocentesis in Beijing and on 16 December 2007 in their home)

Case 3: F and his family feel stigmatised

This case shows how DMD is intimately linked with the family-planning policy and the daily lives of families that carry DMD.

A boy with DMD named F, an 8-year-old primary school student, lives in Hongni village, Wulian County, Shandong Province. His mother, Mrs Y, is 45 years of age. F, who was diagnosed with DMD after taking a genetic test, clearly has problems moving and standing steadily. F has an old sister named X who is 23 and temporarily works in Rizhao city. The doctor advised her to undertake a carrier test. She has already reached the legal age, which is 20, for women to get married in China. F’s father, grandfather and grandmother all did not know clearly about the hereditary character of DMD. Mrs Y had another son who was younger than the daughter. After she gave birth to this boy, she had to undergo sterilization by joining the oviducts [jieza], in agreement with the family-planning policy. Unfortunately, this boy drowned when he was 3 years old. It was a severe shock for the family. After undergoing an operation to ‘unjoint the oviduct’ [shu luan guan fu tong] the mother conceived again and gave birth of F. Now the mother is too old to have another pregnancy. The economic situation of this family that has a boy with DMD in a small village is difficult. Apart from their financial hardships, the mother suffers severely due to self-condemnation and feelings of guilt. Moreover, the family also experience discrimination in the village and feel stigmatized.

32 Amniocentesis is a procedure in which a small sample of amniotic fluid is drawn out of the uterus through a needle inserted into the abdomen. The fluid is then analyzed to detect genetic abnormalities in the fetus or to determine the sex of the fetus.
as a family with a disabled boy. (Interviews with the mother and sister on 9 December 2007 and subsequently)

**Case 4: Ms X and her sons persevere**

This case study draws on information from the Internet and a film, which is based on the true story of a family from Sian, the capital city of Shanxi province. Ms X, a single mother, has twins with DMD named Gold Bean and Silver Bean [Jindou and Yingdou: ‘Bean’ is used as name in Chinese denoting ‘baby’ or ‘sonny’]. Now the boys are adolescents. Ms X divorced when she was 31 and the boys were 6. After her divorce, she brought up and looked after the two affected boys. In order to dedicate herself entirely to the boys and to avoid hurting the boys, she decided not to remarry. Ms Xue strictly asks the boys to take exercise every day, such as a five-kilometre walk. She gives massage to the boys and helps them to do sit-ups every day by pushing their back because they cannot play by themselves. She tried to seek alternative treatment for DMD, which actually was little effective. The boys easily catch flu due to their low resistance, so that she spends much energy and time on caring for the boys. Ms X, who works in a textile mill, therefore cannot afford to work full time. The family lives on her small income and often faces financial difficulties. The experience of the family has attracted public attention, after which their story was written down as a play and was filmed as a television drama named *Love You Using My Life* (yong wo de shengming qu ai ni).

These four cross-sectional cases offer insight into the circumstances and experiences of families with DMD in China. The next sections will address and analyze social issues linked to DMD and DMD genetic knowledge.

**THE SOCIAL AND POLITICAL FRAMEWORK OF CHOICE OF FAMILIES WITH DMD IN CHINA**

Based on the case studies, this section will analyse the vulnerability of families with DMD and related issues such as the social stigma attached to boys with DMD and their families, the lack of welfare support and social help for the families of boys with DMD, and the impacts of
living with boys with DMD on reproductive decision-making and reproductive views.

**Lack of effective treatment**
Twenty years after the discovery of the DMD gene in 1988, an effective medical treatment is still not available (Urtizberea et al. 2003). Although DMD is present from conception, boys with this disorder appear normal at birth. It is when they begin to walk that discrepancies are often noted. In most cases, the child walks with a ‘waddling gait’, tends to tiptoe, and has overdeveloped calves. Later, climbing stairs becomes challenging (Cwik & Brooke 1996). When the symptoms appear, parents initially do not think it is a serious problem and think that the child is just a late developer. Ms L in Case 2 said when she found her son walked slowly she thought it was common that a boy learns to walk later than a girl, or maybe that her son did not take enough calcium and vitamins. She became worried about the ‘walking problem’ after she found her son could not climb stairs like the other boys of the same age. In fact, most families with an affected boy share a similar experience. They come to hospital hoping to get treatment. When parents find out that this is a lethal disorder and that no effective treatment is available, they usually react with feelings of disbelief, denial, anger, anguish and anxiety (DMD Forum 2001). Ms X, the mother of the twins in Case 4, remembers the way she felt:

> I felt like the sky was falling down on me *[tian ta le]*, I nearly lost my consciousness at that moment, like a huge thunderbolt hit my head. The diagnosis result was like a death sentence. I cannot believe my sons will die…. Why am I so unlucky? Why are my sons so unlucky? Why? [Transl. S. Sui]

Ms Y, in Case 3, had a similar reaction:

> I felt so cold. Just like I dropped into an ice cave *[bing kulong]*. My mind became blank *[yipian kongbai]*. If this disease can be cured, I will try to collect money to treat him, even sell the last thing in my home *[zaguo maitie]*. [Transl. S. Sui]
Many families try to find an alternative treatment although the doctors have told them clearly that no medical treatment can cure DMD at this time. They believe what the doctor said but they fervently hope that maybe something else would work. In the clinic, some parents begged the doctor to allow their son to become a human subject in clinical experiments as soon as new therapy for DMD might be developed. Some families turned to traditional Chinese medicine. In fact, according to a pilot study carried out in Beijing on ten boys with DMD treated with traditional Chinese medicine at various stages of their disease course, it is not possible to draw any definitive conclusions regarding the beneficial effect of Chinese traditional herbal medicine in patients with DMD. It seems as if the benefit, if any, is minimal (Urtizberea et al. 2003). Also, parents of boys affected by DMD who are tempted to get access to these drugs often are not informed of their effects and potential hazards, although the potential toxicity of some of these Chinese traditional medicines has clearly been demonstrated elsewhere in other conditions (Chan & Critchley 1996; Critchley et al. 2000). In fact, there are some, usually little known, hospitals that boast in exaggerated advertisements of special treatments for incurable diseases such as DMD (Chen 2005). The advertisements attract people, including those from remote rural areas, whose family member suffers from incurable diseases, such as cancer and DMD. The strong desire for treatment easily misleads DMD-affected families into spending money, worsening their financial conditions. One interviewee in such a situation expressed her feeling of helplessness:

If I do not try, I will feel guilty towards my son. The advertisement said that the treatment will be effective. Maybe there is a possibility. Maybe it will be helpful. I am not rich, but at least I will try once. Anyway, I think that the treatment will not make things worse. I only want to make every possible effort [si ma dang zuo huo ma yi, a literal translation to English is ‘to treat a dead horse like an alive one’]. [Transl. S. Sui].

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Psychological distress and heavy burden for the parents and family

Parents of boys with DMD say they experience depression and emotional anguish. Because some parents view their child as an extension of themselves, they are apt to feel that such a disorder reflects upon them. They feel grief, and a feeling of guilt is a common response to grief. Particularly, mothers with a defective X chromosome have the added burden of knowing that they are most likely the unwitting carriers of the gene responsible for the disorder (Rubin 1987). As the disease progresses, mothers may develop an attitude of self-blame and struggle with guilt regarding the child’s condition. Also, the long-term and escalating nature of DMD creates an increasing burden on families as the disease progresses. Caring for a family member with a severe chronic illness has been associated with increased family stress, diminished health for caretakers and an increased financial burden (Wang & Barnard 2004). Family members must provide increasing assistance with activities of daily life, eventually having to carry them out completely for the affected child. Long-term care needs may increase the financial burden on the family, particularly if one of the parents needs to forego employment to care for the child (Chen & Clark 2007). As a single mother with two sons who need her care, Ms X in Case 4 has a heavy financial burden and psychological stress:

When I was short of money, I only had 20 Yuan (about 2 Euro). I did not know what I could do. At that time I even planned to sell my kidney for money, although I knew it is illegal to sell human organs. Often, I want to cry, shout or sing loudly. I even considered committing suicide with my sons, so as to finish it all [yì liáo bài liào]. Sometimes I cannot sleep at night and stand in the dark, smoking cigarette after cigarette. In that year, when my sons were 12 years old, I started to lose my hair. I shed a lot every day. In addition, every day I accompanied my sons for five-kilometre walks and I engaged in massage and stretching exercises for my sons. I often feel my body and soul are all exhausted [xīnlì jiāocuí]. [Transl. S. Sui].
Ms Y, in Case 3, also feels she is under the pressure from guilt, depression and anxiety:

I feel life is black and colourless. Watching my son walking in that way, thinking he will be unable to walk and that he will die, my heart hurts so much, like it is being pricked by a sharp needle [xin ru zhen za]. If I could, I would like to suffer instead of my son [she said crying]. It is my fault, and I feel so bad and feel so sorry [dui bu zhu] for my son and my husband. If my husband wants to divorce me, I have nothing to say. My son is still young and he does not understand his disease, and I do not want him to know more. I only hope he can be happy. [Transl. S. Sui].

From Ms X’s and Ms Y’s narratives we can understand the heavy psychological burden of the mothers.

**Stigma and discrimination against families with Duchenne muscular dystrophy**

Ervin Goffman, a well-known sociologist, defined the concept of stigma as denoting an attribute that is deeply discrediting that reduces the possessor in our minds from a whole and usual person to a tainted, discounted one (Latz 1981). The attribute that stigmatizes one type of possessor can confirm the usualness of another, and therefore is neither creditable nor discreditable as a thing in itself (Goffman 1986:3). The ‘abomination of the body’ is a type of stigma, consisting of various physical deformities, disabilities and chronic diseases. People use specific stigma terms such as cripple and moron in daily discourse as a source of metaphor and imagery (Goffman 1986:5). In practice, persons suffering from a severe illness and their close family are often socially stigmatized (Sartorius 1997). A popular view in China considers severe illness, to a certain extent, to be punishment for an ancestor’s misbehaviour or for the family’s current misconduct, which is called ‘religious punishment’ [zongjiao chengfa] or ‘pre-existence retribution’ [qianshi baoying] (Lin & Lin 1980; Wang & Zhang 2002; Liu 2006). This view, which is a popular explanation of pathogenesis based on religion and superstition, sometimes is used to stigmatize families with severe handicaps, causing conflict and dissatisfaction. Such stigma and discrimination make the disabled feel
distressed. In fact, it is very difficult for the disabled to lead a ‘normal’ life, so that it is challenging for the disabled to get married, and the disability sets them apart from their friends and neighbours (Kohrman 1999). As a lethal inherited disorder, DMD instils fear into people. The long process of progressive weakening of the patient, and the late-onset genetic nature of the disease resulting in early death, intensifies the stigmatization of the family. Stigma and discrimination teach family members self-contempt and guilt, isolating them from the community. The DMD-affected family members interviewed all felt that their family was suffering from stigma and discrimination. Mr S, the elder brother of Ms S in Case1, said his childhood was full of painful memories of having an affected young brother:

Sometimes several boys in the neighbourhood followed him, simulating his walking style, mocking him, and calling him ‘crippler’ [que zi]. This illness is monstrous and strange. One of my uncles died from it. My family was the target of gossip in the community, which enjoyed chatting about my family. Some words, like ‘misconduct’, ‘sin’, ‘retribution’ and so on [zuo nie, zui guo, bao ying] would eventually reach the ears of my family. My family could not stop it and dared not to do anything because we were afraid that the gossip would worsen.

I am so lucky to be the one who is unaffected. Now, I have drawn a pedigree and found those who are at risk of being a carrier and developing the disease. I will try to contact them and offer them money for them to take a test. I hope no newly affected boys and no new carriers will be born to my family. Let this damned illness disappear from my family. [Transl. S. Sui].

Ms L, in Case 2, and her husband are also afraid of stigma and discrimination:

The symptoms of my son are getting obvious and we cannot keep it secret, but we do not say more about this illness to others except to our very close relatives. We are afraid of gossip. We are very cautious and scrupulous to avoid offending anyone. In school, several classmates laughed at my son and he feels bad. The only thing we can do is to implore the teacher to pay more attention. [Transl. S. Sui].
Ms Y also feels that she and her family are severely stigmatized:

We cannot hold our heads high in the village. We lost one son, and we will lose this one. A family without a son is despised in this village. Some people think our family is cursed by a devil. In our village, usually, a matchmaker will come to a girl’s home to propose a candidate for marriage [ti qin] when she approaches the age of 20. My daughter is 23, and no matchmaker came for her. [Transl. S. Sui].

The girl, the daughter of Ms Y, expressed her feeling about this:

I feel pity for [kelian] my little brother and my mother. I do not want others to know about the situation of my little brother and my carrier state. I keep it secret in the place where I work. I have no boyfriend at present. If I have a boyfriend, I do not know whether I will tell him my carrier state or not. Maybe I will tell him. I think I should not cheat and it cannot be kept secret for ever. Many of my classmates are already married. You know, in our small place early marriage is common. Now I do not think more about this. What will be will be [shun qi ziran]. [Transl. S. Sui].

One interviewee in this study, a carrier of DMD, also narrated her experience. She said that the parents of her boyfriend strongly set themselves against the relationship between her and their son when they found out about her carrier status, and finally her boyfriend separated from her. This case showed the severe stigmatization of the DMD-affected family, worsened by popular superstition.

**Implication for reproductive decision-making**

Gender preference in Chinese views on reproduction is not unusual, and expresses a strong male bias (Zheng 2004). This preference is expressed in the burning of incense for ancestors,
a revived tradition increasingly prevalent in China. In Chinese traditional reproductive views, only the male is treated as family offspring, which is called ‘burning incense’ [xiang huo: in Chinese it means the son/sons in a family] (Liu 2005), for traditional custom prescribes that only a son can hold a memorial ceremony for the ancestors. So, ‘burning incense’ used in Chinese especially means the male offspring of a family. However, because boys are symptomatic for DMD, the DMD-affected families have strong preference to have a girl rather than a boy. Ms S’s words in Case 1 illustrate this:

If only this fetus were female, I could unburden my mind and relax. But I already know he is a boy. If I am a carrier, there is a 50 per cent risk that he will be affected. I cannot or dare not take such a high risk. You know I hurt a lot. My baby is already a little person now. I can feel him moving every day. It is so great to feel his action. I am such a cruel mama. I really hope that my baby is a girl; if it were, nobody could persuade me to give her up. [Transl. S. Sui].

And Ms X, mother of the twins with DMD:

My family was very proud of the birth of my twin sons before the boys developed symptoms. When the boys were 1 month old [man yue], their grandmother boiled two hundred eggs and coloured them red after which they offered two to every family in the community. Our neighbours and friends all admired me very much. But now, they all feel sorry for me. Sometimes I speculate about what if my twin sons were twin girls, or even just one girl…. But life cannot be changed. [Transl. S. Sui].

Ms L expressed her joy when she found out that her fetus was a girl:

When I had just conceived this time, I always prayed. I hoped that God would give me a girl. I also had a strong feeling this fetus was a girl. I am really happy to know she is a girl. I do not care if she is a carrier or not. [Transl. S. Sui].
It is no doubt that boy-preference is the major factor influencing the Chinese couple’s expectation and action for reproduction. In some rural areas, sex-selective abortion is also the main reason for the skewed sex ratio in China, particularly in rural areas (Chen 2004). In China, one of the main issues in birth policy-making is that many second children were allowed as exceptions to the one-child ideal. The upshot was that about half of the rural couples whose first child was a daughter were allowed to try again for a son. This birth policy in rural areas remains in effect in the early twenty-first century, subject to informal debate, but is evidently not yet time for formal reconsideration (Greenhalgh & Winckler 2005). The exceptions in the one-child policy imply and represent the son-preference in China. But, in the context of DMD, sex selection is reversed.

**Social isolation of the affected boys**
Children with disabilities are limited in their activities in society. According to research on issues of the disabled in China there are an estimated 2.15 million children with a disability who are not receiving schooling. However, 76 per cent would be physically able to attend school, but have no opportunity to do so. Furthermore, only 5 per cent of young people with a disability live in the cities, where they might attend special schools for the disabled, which are scarce services in China. This leaves the majority, who live in the countryside, uncatered for (Stratford & Ng 2000). Similarly, children with DMD in this study have little opportunity to join their peer group in school, because of walking difficulties, which make them more likely to be socially isolated.

This research on DMD-affected families suggests that they also experience great social isolation from the community. For instance, parents report that their boy does not like going to school and wants to stay at home to watch TV instead. Ms Y complained:

> My son spends much more time watching TV. Sometimes he sits before the TV set for a long time, even though the programmes are not interesting. I think he is getting more sensitive as he is getting older. He is afraid to get hurt outside and tries to avoid it.
The father in Case 2 also reported that his son had become quiet:

My son does not like to play with other children even when they invite him and are friendly to him. He likes watching TV, and sometimes draws pictures. I think that he feels inferior when he plays with other children. You know, he cannot run or even walk properly.

Examples of the social isolation of boys with DMD show that such boys feel unsafe and feel they are not accepted by the community.

Support from society

In China, there are several Internet communities for the ‘muscle illness patient’ [ji ji huazhe]. These Internet communities, such as ‘Going with you’ [jingcai tongxing] and ‘window for muscle atrophy’ [ji weisuo zhi chuang] offer virtual space for patients and their family to communicate and share their experience. Also there are several websites that offer general genetic knowledge and answer questions online, which is called ‘counselling on-line’ [zaixian zixun], such as ‘muscle atrophy web’ [ji weisuo wang] and ‘genetic question and answer web’ [yichuan wenda wang]. The resources provided through the Internet to some extent are helpful to patients with DMD and their families. But in China, most rural areas and small towns have no access to the Internet. And to most villagers the Internet is far away from their lives, as they do not know how to use a computer in the first place.

In China, in December 2005, the Red Cross established the DMD Fund, called ‘kindness from an angel’ [tianshi zhi en] (Zhang 2005). The purpose of the fund is to help boys affected by DMD. At its launch, the fund valued 100 thousand Renminbi (about 10 thousand Euros). It had been donated by a video company and contributed to DMD-affected

38 Address of the website: http://www.genet.org.
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twins Gold Bean and Silver Bean, on whose case the film Love You Using My Life [yong wo de shengming qu ai ni] was modelled. But afterwards, the fund was only able to help one boy with DMD in Beijing with a donation of ten thousand Renminbi (about 1,000 Euros) in 2007 (CRCF, 2007). None of the DMD-affected families involved in this study had heard of the fund, and they did not regard applying to it as a viable option.

CONCLUSION

Based on the cases of the four of DMD-affected family and their narration of personal experience and feeling, this paper focuses on the socio-economic aspects of the DMD-affected family and analyzes related social issues in Chinese contexts, such as reproductive decision-making, family stigma and social discrimination, and the difficulties the family affected by DMD encountered in the Chinese contexts. In current China, DMD-affected families are not only undergoing economic hardships, which are caused by the cost of ineffective treatment and energy and time spent on the attentive assistance for children affected by DMD, but also facing heavy psychological pressure as a result of family stigma mainly coming from the view of ‘pre-existence retribution’ as a explanation of pathogenesis based on religion and superstition. Meanwhile, the Chinese cultural values attached to healthy offspring increase their psychological burden, especially for the mothers, who usually are strongly self-condemned and are blamed by family members. Families affected by DMD are vulnerable, and the existing social discrimination aggravates their vulnerability. Additionally, social welfare and support, due to the lack of accessibility and effect, do not play a full role in reducing the vulnerability of these families.

Other issues that should be explored are the present non-professional modes of genetic counselling and its implications in Chinese contexts. Therefore, more social-scientific research is needed to understand the application of genetic technology and the social implications in this field.