1-1-2003

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Bachelor of Arts
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1989

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A thesis submitted in partial fulfillment
of the requirements for the

Master of Arts Degree in Psychology
Department of Psychology
College of Liberal Arts

Graduate College
University of Nevada, Las Vegas
May 2004

UNLV
UNIVERSITY OF NEVADA LAS VEGAS

Thesis Approval

The Graduate College
University of Nevada, Las Vegas

March 30, 2004

The Thesis prepared by

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Entitled

The Psychological Impact of Carrier Status in Hereditary Disorders

is approved in partial fulfillment of the requirements for the degree of

Master of Arts in Psychology

Examination Committee Chair

Dean of the Graduate College

Examination Committee Member

Examination Committee Member

Graduate College Faculty Representative

1017-53

ABSTRACT

**The Psychological Impact of Carrier Status
In Hereditary Disorders**

by

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Previous studies have indicated that carrier status has little effect on self-concept. However, rather than examine autosomal recessive illnesses, wherein genetic responsibility is shared by both parents, the present study samples women at risk of being sole carriers of an X-linked hereditary disorder, Duchenne muscular dystrophy. Previous research most often assessed global self-esteem, thus problems in particular domains (such as future parental role or desirability as a mate) have generally been overlooked. Herein, a stigmatizing process is hypothesized whereby one aspect of the self, genetic identity, may be spoiled through a diminished sense of worthiness to reproduce. Family attitudes toward risk, how important bearing "her own" biological children is to the woman, how many social roles she currently enacts, whether she has had genetic testing or genetic counseling, and whether she has utilized follow-up counseling to aid in coming to terms with her carrier status, may moderate stigmatization.

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ACKNOWLEDGMENTS

My plan for the line of research embarked on in this Master's thesis began underground many years ago. It surfaced in August, 2002, thanks in large part to the insistence of teacher/trainer extraordinaire, Sylvia J. Mills, that I identify 'my dream.' At UNLV, Dr. Murray Millar generously agreed to take over mentorship as we framed my question as a health behavior, social psychological inquiry. (Dr. Michael Hall had mentored me through a year of music perception and cognition studies and another of engineering psychology centered around the effects of cell phone use upon drivers' attention and auditory perception. He graciously assisted me in finding a new mentor when we came to a fork in the road.) In addition, I thank Dr. Allyn McConkie-Rosell of Duke University for valuable input and encouragement. My committee members Dr. Daniel Allen and Dr. Mark Floyd of Psychology and Dr. Daniel Benyshek of Anthropology offered their time, helpful suggestions, and cheerful support.

CHAPTER 1

INTRODUCTION AND LITERATURE REVIEW

The present study proposes that women at risk for passing on an X-linked defect (specifically, Duchenne muscular dystrophy) experience stigmatization in the form of a spoiled genetic identity through a diminished sense of worthiness to reproduce. It will investigate, as possible mediating variables, a diminished sense of desirability as a mate or potential mate and as a parent or potential parent. Further, a number of variables that may moderate the impact of carrier status on perceived self-worth will be examined. Variables proposed as moderating the degree of stigma experienced include input from family of origin, the personal importance of biological children, the number of concurrently active social roles, as well as the type and extent of counseling received.

Psychosocial Implications of Genetic Testing and Screening

As knowledge of the human genome increases, genetic testing is becoming possible for an ever greater number of hereditary disorders. When such tests are run as a matter of course either among the general population or particular subgroups considered at heightened risk, testing becomes part of a broader approach known as genetic screening. Marteau (1992) has pointed out that for many individuals, genetic screening programs will mean finding out for the first time that abnormal genes are carried by almost everyone.

Although carrying a recessive gene only very rarely compromises the health of the carrier, Fanos and Johnson (1995) found that 30% of cystic fibrosis siblings assumed that being a carrier conferred health problems. In another study, when followed up one to three years after testing, persons found to be carriers of Tay-Sachs disease had a more pessimistic view of their future health than noncarriers (Marteau, van Duijn, & Ellis, 1992). With regard to sickle cell anemia, 40% of noncarriers and 20% of carriers studied by Stamatoyannopoulos considered carrier status ("sickle cell trait") to be a mild disease (1974). Not only is the actual determination of a risk factor often problematic due to indeterminate testing technology, but the meaning of a particular probability varies greatly from one individual to another. Irrespective of income and education level, misconceptions, family folklore, and superstitious beliefs often govern potential carriers' viewpoints (Fanos & Johnson, 1995; Kay & Kingston, 2002).

The psychosocial complications of undergoing genetic testing and the resultant knowledge of one's own genome include possible stigmatization. Markel (1992) observed that, historically, the perception of groups as genetically inferior is accompanied by social ostracism and stigmatization. Immigration to the United States, for example, by Jews, Italians, Greeks and Balkans was drastically reduced by the Immigration Exclusion Act of 1924 largely due to the efforts of Charles B. Davenport and Harry H. Laughlin of the Eugenics Record Office at Cold Spring Harbor, New York. Their propaganda centered on the misguided idea that these groups represented a threat to the American gene pool because of traits ascribed to them such as poverty and lust. A similar process can occur regarding bona fide genetic factors, as demonstrated by the results of a screening program for sickle cell anemia in Orchomenos, Greece. In the small

farming community, the families of persons who were identified as carriers saw avoiding matches of two heterozygotes as an additional complication to courtship that resulted in reduced freedom in making marriage arrangements. Among couples who married following the screening program 20% to 26% equated sickle cell trait (the carrier state, referred to as AS genotype) with risk of social stigmatization (Stamatoyannopoulos, 1974).

The large scale screening effort regarding sickle cell anemia among African Americans in the 1970s exemplified the negative social implications that can arise. Assuming that prevention of the birth of affected children is the goal, reducing the size of the community receiving the screening and counseling is the inherent result. When a genetic defect is prevalent in members of a particular ethnic group, that result could be viewed as a form of genocide (Stamatoyannopoulos, 1974; Whitten, 1973). When ethnic concentration of a genetic trait is not at issue, the goal of prevention creates an individual analog to genocide, the termination of that person's genetic line. Key to such interpretations, however, is viewing childlessness as the obligatory course for carriers. Options increase with *in vitro* fertilization in conjunction with preimplantation diagnosis, early prenatal testing, or sex selection techniques in which carriers can opt to either conceive or birth only unaffected offspring.

Psychological effect of stigmatization. In 1989, Wooldridge and Murray used a self-report scale that they had developed to assess feelings about sickle cell trait (carrier status). Their Health Orientation Scale is a semantic differential self-report regarding nine hypothetical scenarios. They found that both carriers and noncarriers had very positive feelings about themselves in general, but that noncarriers imputed more negative feelings

to carrier status than did carriers themselves. The latter provides evidence of social stigmatization. When considering whether to tell friends of their carrier status, carriers' descriptions of their own feelings dropped from positive toward "marked." When contemplating the risk to potential children, carriers' descriptions of their own feelings dropped toward "marked" and "guilty". Such variance supports the concept that damaged self-concept (spoiled genetic identity) is experienced particularly in regard to reproduction and self-disclosure.

Jones et al. (1984) proposed that individuals construct a sense of self based on the affective reactions of others. Through interaction with others and learning about ourselves from their reactions to us, we develop a concept of a self distinguished by abilities, achievements, preferences, appearance, and temperament. Self schemas are viewed as the relatively enduring, strong views one has about the self. The stigmatization process starts with the existence of a "mark," a characteristic or feature that has been labeled as discrediting by the surrounding social milieu, sometimes implicitly due to assumptions so basic that they are seldom consciously acknowledged. In such cases, individuals may "self-stigmatize" through having internalized the implicit assumptions of their social framework. However, to move toward stigmatization, the person must attend to the mark. Here, the messages inculcated by family of origin play a major role in determining how salient the mark appears to be to the marked person. In the case of physical handicaps, for example, the responses "you can't do that" and "we'll find a way" will contribute to quite different levels of a child's attention to a disability.

When great attention is paid to a mark over a prolonged period, the marked individual is likely to organize the self around it. The mark and the sense of being

discreditable become, at that point, incorporated into a self-schema and form a focal point of the self-concept, leading to spoiled identity (Jones et al., 1984). When the mark is of a genetic nature, it is one's genetic identity that is spoiled. That is, genetic defect discredits the person's permanent, unchangeable, inherited traits. In as much as it is through offspring living into the future when the current generation will not, genetic identity "represents the sense of immortality evidenced in the continuation of family blood lines" (Schild & Black, 1984, p. 54). When continuation of the family blood lines is endangered due either to premature death of the offspring or through lack of reproduction, genetic identity is challenged.

Stigma always occurs within a social context (Goffman, 1963) as it requires a difference that is socially selected for attention, labeling, and negative evaluation (Link & Phelan, 2001). The social context may be time-dependent in that differences may have greater social significance at particular life stages than at others. Dating, courtship, and marriage occur during a time period during which desirability as a mate is of greatest importance. It may only be within that particular life stage that the elements that result in stigma are manifest. Link and Phelan identify four components of stigma: the distinguishing and labeling of a difference; linking persons so labeled with undesirable attributes that are part of a negative stereotype; separating the marked person as fundamentally different ("us" and "them"); and the different person losing status and being discriminated against by means of greater social power being held by those who devalue the trait. Similarly, Berger, Ferrans, & Lashley (2001) propose that prior to perceiving stigma, persons must be aware of societal attitudes toward a trait and know that they themselves possess the trait. If, further, they are aware that such attitudes result

in "social disqualification, limited opportunities, or a negative change in their social identity (p. 520)," then they perceive stigma. Berger et al. add that although some may respond to perceiving stigma with a negative change in self-concept, others will challenge the stigmatizers, react emotionally against them, or use avoidance or humor to minimize stigma.

In the case of women carrying a deleterious gene, genetic difference has become more distinguishable with the development of genetic testing. Fear of bearing or begetting an affected child derives from familiarity with the discrimination long experienced by the disabled and the courtesy stigma often experienced by those associated with them. The prospect of avoiding children, however, leaves one open to yet another stigma, that of the "barren woman" or, in Latin America, "yerma" or "arid desert" (Acero, 2004).

Mediator

The mechanism through which stigmatization and loss of self-worth occurs in the case of genetic carriers is proposed to be through a sense of lessened desirability as mate and future parent. The process involved has both psychological and social aspects. Psychologically, the awareness that one cannot or might not produce a normal child implies that one is inherently flawed and deficient and can be experienced as "a narcissistic wound, i.e. as an attack on the self-system" (Kessler, Kessler, & Ward, 1984, p. 679). Since the area in which the carrier would be deficient is that of producing children, and offspring are a generally assumed result of mating, carrier status necessarily compromises the carrier's desirability as a mate.

Socially, the more essential childbearing is to a woman's social status, the more damaging carrier status will be. In many cultures where arranged marriage is the norm, identification as a carrier of a genetic disease has clear disadvantages in the marriage marketplace. Regardless of locale, however, the consequences of foregoing children due to genetic risk add further blows to self-worth, as childless couples are frequently viewed as deviant and subject to negative stereotyping. As Veevers concluded in 1972, the social interpretation of nonparenthood can include being irresponsible, immoral, unnatural; lacking sexual competence, rejecting gender roles, threatening marital stability; displaying immaturity and emotional maladjustment (as cited in Hollerbach, 1979). A woman aware, consciously or unconsciously, of such repercussions may feel as one woman described her experience with infertility to Jill Eisen: "I felt guilt, lots of guilt. I felt that I had stuck my husband with this woman that would never give him the children that we had wanted. I felt asexual, I felt very neutered by the whole experience. I felt I had lost all my womanness" (cited in Overall, 1987).

In Fanos and Puck's study (2001) of siblings of boys with X-linked severe combined immunodeficiency (XSCID), sisters reported perceiving themselves as less desirable as mates and feared that knowledge of their carrier status would eliminate their chances of finding a partner. Complicating matters, potential carriers often feel obligated to anticipate being rejected. In their interviews with 14 identified carriers of X-linked disease, Kay and Kingston (2002) found that many felt a responsibility to disclose their carrier status "early on in the relationship." They felt that the mark should be revealed in the expectation that it would likely make them undesirable, at least to some suitors, "giving them a chance to get out if they were scared" (p. 175). The belief that it is not

fair, or not responsible, to conceal the marked condition is often inculcated in young women as they are growing up, based upon the concept that the mark would discredit, or make her less acceptable, to a future partner, and that it would be dishonest to misrepresent oneself as not having the mark. The more discreditable carrier status is felt to be, the earlier in a relationship one may feel obliged to disclose her situation.

That suitability as a parent is involved is clear from the widespread guilt felt by women who bear a disabled child. They feel diminished, discredited as a parent. A frequent reaction is to have no more children, as if they felt disqualified by "the potential for a defect in one's products" (Kessler, Kessler, & Ward, 1984, p. 679).

Moderators

A number of variables may moderate the amount of stigmatization that accompanies carrier status. Moderating variables include input from family members, the importance of biological parenting to the individual, the number of roles being enacted, genetic testing and counseling, and follow-up counseling.

Input from family. The family's influence on the development of genetic identity is paramount because it is generally only within the family that one's genetic makeup is known, that carrier issues are discussed. This input is likely to be biased against reproduction, coming as it often does from individuals whose offspring did inherit genetic disease. They played the odds and lost, thus know firsthand the answers to the "what ifs." Not surprisingly, they often have a negative bias toward 'risking it.' This negativity may extend to childrearing in general. The more family members who communicate negative messages to a young woman, the more diminished her sense of

worthiness as a mate and parent is likely to be, hence the greater the stigma she will experience.

Women that have had children, even when none of the children inherited the genetic disease, are likely to recommend carrier testing at an early age (preteen years, at least by age 12) (McConkie-Rosell, Spiridigliozzi, Iafolla, Tarleton, & Lachiewicz, 1997). While they feel that they would have avoided biological children had they known the risks, their daughters who avoid bearing children may come to feel that their options were unfairly reduced before they could determine them for themselves. Especially where testing currently yields inconclusive negatives, or false positives, delaying carrier testing until childbearing age may allow time for better technology to develop.

For example, Kelly (1977) reported the case of a woman whose risk factor for oculocutaneous albinism was 1:600. But the woman's mother (who had accepted a 1:4 risk in bearing her own children) insisted that her daughter's risk was 50/50, and made stinging remarks conveying her opinion that it was too high a risk for her daughter to consider accepting. It is worth noting that there is a tendency to jump from there being known risks in pregnancy to the idea that the young woman "should not have children." Oftentimes other options, such as sex selection, prenatal diagnosis (with the possibility of termination of affected fetus), or even adoption are not acknowledged. At times carrier testing is not even performed, a high risk being a foregone conclusion. For example, Dorothy A. remained unmarried past the age of 33 because of an offhand remark made to her during her teens that she should never reproduce, her two brothers being crippled by a muscle disorder. Struggling to reconcile herself to remaining single and childless had

caused her such emotional trauma that she sought help and was finally referred for carrier testing by her psychiatrist (McCollum & Silverberg, 1979).

Females may be particularly vulnerable to family constructions of risk and resultant expectations. Gilligan (1982) described an "ethic of care" and emphasis on relationships and connectedness among women in contrast to an emphasis on autonomy among men. (For a review of related literature see Statham & Rhoades, 2001.) Daughters, then, may find it especially difficult to go against familial bias against "taking the risk" of pregnancy, for fear of rupturing bonds with those having the most inflated perception of risk and therefore the most opposition to pregnancy. At the same time, however, the role model provided by most mothers of disabled children is likely to have been that of a full-time homemaker. The result is a double-bind situation of "Do as I say, not as I did."

Even in the case of autosomal recessive cystic fibrosis, it has been found that can family history can override rational assessment of risk. Denayer, Welkenhuysen, Evers-Kiebooms, Cassiman, and Van den Berghe (1996) found that of the carriers interviewed who had personal experience of the death of a sibling with CF, some decided to have no children or seemed convinced that their children would have CF (irrespective of whether their future partner was a carrier). Others felt that having a CF child would be an expression of their own conviction that the life of their sibling was not meaningless.

Although considerable research on psychosocial impact of genetic testing and carrier status has focused on autosomal recessive traits (e.g., Tay-Sachs, cystic fibrosis, sickle cell anemia), one X-linked disease that has been the subject of investigation is fragile X syndrome, a common cause of mental retardation. Research with this population sheds light on the extent to which parents wish to influence daughters' choices out of

concern for their welfare. In interviews with 28 women who were tested because of their affected child and were found to be carriers, McConkie-Rosell and her associates found that 93% felt it would be preferable for young women to know that they are carriers before entering serious relationships (McConkie-Rosell et al., 1997). The reasons given were either to be able to make a mutual, informed decision about reproductive risk or to "marry someone willing to not have children" (p. 65).

Most women felt that their own unaffected children should be tested at ages as young as 4 years old. However, women who reported being relatively more upset by their own carrier status and viewing themselves more negatively since learning of it, preferred to 'test and tell' once their children were over 16 years of age. On the other hand, women who saw younger ages as appropriate for testing and knowing results viewed the risk of affected offspring as higher than those who would wait until their children were older. Parents of children with fragile X reported in another study (McConkie-Rosell et al., 1999) that their motive for learning their child's carrier status was not merely to relieve their own anxiety, but in order to "provide anticipatory guidance" to help the child adjust to carrier information. Viewing such guidance as a parental duty, they were convinced that the right to test minor children rightfully resided with parents. Mothers were more concerned than fathers that children be informed early, before the possibility of bearing or begetting children. A major element was interest in preventing the child's later resentment for not being informed, were they to bear an affected child. It must be kept in mind that the perspective of parents in both studies is necessarily affected by the psychosocial impact of having borne affected children without advance suspicion of

carrier status. Such a perspective thus possibly ignores the negative complications of early knowledge of carrier status.

Importance of biological parenting. It has been suggested by McConkie and DeVillis (2000) that the aspect of self-concept negatively affected by suspected carrier status is that of future parental role. Having carrier status confirmed by genetic testing does not necessarily close the door on pregnancy and childbearing. There are, after all, the options of prenatal testing which has always carried the implication that affected fetuses will be terminated (Cowan, 1992) and *in vitro* fertilization with implantation of only females, or unaffected males (The Reproductive Specialty Center, 2003). Either alternative is costly, in emotional or financial terms, or both. However, some physicians profess to have developed guidelines to encourage the natural conception of one sex or the other (Shettles & Rorvik, 1997; Whelan, 1991) and sperm sorting wherein the father's ejaculate is centrifuged to separate X-carrying from Y-carrying sperm (The Reproductive Specialty Center, 2003). The X-carrying sperm are used to artificially inseminate the mother, assuring the conception of daughters only (MicroSort, 2003). In spite of these alternatives, there seems to be a frequent leap from carrier status to "not able to have children," perhaps out of ignorance or unacceptability of alternatives or as an outgrowth of spoiled genetic identity. It would be expected that the greater the importance of biological parenthood to an individual woman, the more vulnerable to stigma she would be.

Low desire for biological parenting could simply be an individual preference, or it could signal a denial of something perceived as impossible to attain, an attitude adjustment serving as a coping mechanism. Attitude change from wanting to bear

children to not wanting to do so, would reduce cognitive dissonance stemming from the perception of "not being able" to have children. Depending upon a person's age when carrier status is first considered to be a real possibility, such a coping mechanism may predate any development of desire for offspring. It may also result from influence of family attitudes (to be discussed later) regarding the culpability of those bearing an affected child (thus the unacceptability of any level of risk) or toward other options such as adopting children.

The importance of the role of biological parent, or future biological parent, in a woman's hierarchy of identities can be described by the role's salience and her commitment to it. Stryker and Serpe (1982) define *salience* as the likelihood that an identity will be claimed in a variety of situations and *commitment* as the degree to which her relationships to various groups of others depends upon her playing that role. Commitment can also be viewed as amount of time and energy invested in the role (Marks, 1977).

Multiple roles. According to Goffman (1963), stigmatization is a product of the individual's social situation. A concealable defect would therefore socially stigmatize one only when in the company of those to whom the carrier feels obligated to divulge the information. One means of managing stigma, therefore, would be to increase one's social circles in which the mark is irrelevant and remains hidden. That is the basis for viewing multiple roles as a moderator of diminished self-worth due to carrier status. The more interaction the marked individual has with persons or groups to whom they are "normal" (in whose company their genetic identity does not affect their social identity), the less significant the mark may seem and the less the self would be organized around it. Since

the stigma attached to carrier status is "visible" only in situations where childbearing is relevant, women who function in areas unrelated to childbearing should experience less stigmatization.

Multiple roles would facilitate engagement in arenas where high achievement is possible, offsetting that where it is perceived to be unlikely (parenthood). Hence, if alternate social roles are being enacted, the perception of stigma resulting from problems in one role should be less than that for women who do not have non-maternal roles.

Identity theory (Stryker & Serpe, 1982) defines the self in terms of occupied social positions. Thoits (1983) stipulates that to be protective, structural positions need to be accepted and enacted through roles that are personally meaningful to the individual. She further proposes through the "identity accumulation hypothesis" that the more identities held through enacted role relationships, the greater "purpose, meaning, direction, and guidance to one's life" (p. 175). Having a meaningful career, for example, may lessen distress associated with not feeling free to have desired children. Assuming this is the case, the relative importance to the potential carrier of being a parent, particularly a biological parent, is another important moderator of diminished self-worth (stigma).

Genetic testing. Accurate information relating to a potential carrier's status is the starting point in promoting healthful attitudes among carriers. Much of the distress surrounding reproductive issues involves dealing with uncertainty as to one's carrier status. DNA testing with PCR (polymerase chain reaction) or Southern blot analysis can reveal the presence of 98% of possible deletions in the dystrophin gene, many of which cause shifts in the reading frame of the three-amino-acid codons that encode protein

synthesis. However, at least one third of DMD cases are caused by small ("point") mutations that are not detectable by these methods (Amalfitano, Rafael, & Chamberlain, 1997). Thus, female relatives of such patients are unable to receive a definitive identification, particularly assurance that they are *not* carriers. Another complication occurs when the DMD patient is deceased and no DNA is available to ascertain the particular deletion or mutation responsible for the disease in his case. Tests do exist that can detect virtually all carriers of DMD (e.g., denaturing gradient gel electrophoresis, DGGE), whether there is a family history of the disease or not (Dolinsky, de Moura-Neto, & Falcão-Conceição, 2002). However, given the lag time from new applications of technology to wide-spread use, uncertainty is a problem that will likely continue for some time. The complexity of carrier status determination is often underappreciated, not only by family members, but by physicians as well, underlining the value of consultation with specialists, that is, genetic counselors or geneticists.

Shame-based perceptions are frequently dispelled to some extent by facts. Since concern about carrier status frequently predates testing, those whose concern is validated will likely feel no further decrease in self-worth, while those who find their risk minimized may experience relief and an enhanced sense self-worth. The overall effect would be toward less sense of stigmatization.

Genetic counseling. In addition to accurate diagnosis, assistance in interpreting the results of genetic testing is essential. On one level, test results must be considered in conjunction with family pedigree and whether the affected family member is available for comparison testing, the individual's willingness to accept the reported degree of risk, and the familial attitudes to which they have been exposed. On another level, given a definite

or possible carrier status, assistance in identifying available options is helpful in moving from the stigmatizing "You're a carrier, you can't have children" to "You can have children, should you wish to, by minimizing or eliminating risk of DMD, through any of the following strategies."

Lower scores on stigma-related items will be associated with (1) obtaining accurate information (genetic testing) and (2) communication regarding the limitations of the test and a risk factor that takes other information, such as family pedigree, into account, as in Bayesian analysis (formal sessions with a genetic counselor).

Follow-up counseling. Furthermore, the degree of stigma should decrease with (3) discussion of one's emotional reaction with a trained counselor or therapist, individually or with a group, given that the goal of such counseling is reduction of isolation; assistance in recognition and acceptance of feelings toward the disease, personal risk, and life goals; and assessment of options. By moving the family skeletons from the closet out into the light of day, they can be deprived of their power to frighten, intimidate, and stigmatize.

Choice of X-Linked Condition

To examine such effects, potential carriers of the X-linked condition Duchenne muscular dystrophy (DMD) will be used as a model. This choice is based on the rationale that implications for self-worth (effects on the self) are more distinct when carrier status resides in one individual, rather than when inheritance depends on both parents being carriers. The majority of the few studies that address feelings of stigma among carriers have centered on autosomal recessive conditions (e.g., Denayer et al., 1996; Evers-Kiebooms, Denayer, Welkenhuysen, Cassiman, & Van den Berghe, 1994, cystic fibrosis;

Massarik & Kaback, 1981, Tay-Sachs disease). To carry an autosomal recessive genetic trait typically does not result in passing on the illness itself, unless the other parent also happens to be a similar carrier. Exceptions would be when a child inherits carrier status of codominant diseases such as Sickle cell anemia. In such cases possessors of Sickle Cell Trait (i.e., carriers) can suffer some symptoms. In X-linked illnesses rare "manifesting carriers" also exhibit some symptoms. In autosomal recessive defects, the 'burden of responsibility' is shared, rather than being focused on one person. One carrier of cystic fibrosis explained that it was problematic only until ascertaining that his future partner was not also a carrier, then it had negligible importance (Denayer et al., 1996). In other words, there is nothing in either of the prospective parents that would make their unique contribution inherently damaging. Only when paired with another of like genetic makeup do carriers of autosomal recessive conditions pass on the actual illness. In contrast, no matter with whom an X-linked carrier mates, the risk is present; the risk for passing on this particular disorder resides solely with her. Thus, in choosing an X-linked disease as a model, we can better examine the stigmatizing effect of carrier status.

Another reason for using carriers of X-linked disease in the study of related stigma is that they are, by definition, all female. There is evidence even with regard to autosomal recessive conditions, that females are more negatively affected by carrier status than are males (Evers-Kiebooms et al., 1994). Whether carrier or non-carrier, females expressed stronger feelings about carrier status than did men. This makes intuitive good sense, as women are the biological carriers of the fetus, hence viewed as having primary responsibility for its health. Such an assumption would be far more reasonable in terms of prenatal nutrition than of genetic endowment. But in the face of

societal readiness to 'blame Mother,' the inclusion of fetal genetics within women's area of responsibility seems ingrained, if nonsensical.

The prognosis of many X-linked diseases is dismal. The lethality and great burden of care associated with DMD is another reason that carriers of this disease bring into distinct relief the extraordinary implications of child-bearing and the possibility of associated stigma. Duchenne muscular dystrophy is a progressive, lethal disorder resulting from defects in the dystrophin gene which is located on the short arm of the X chromosome (at Xp21.1). Due to nonfunctional or absent dystrophin molecules in skeletal muscle, the muscle membrane is inadequately attached to the contractile apparatus in the muscle fiber. Membrane damage ensues, with subsequent disruption of biochemical processes. Eventually damage to the cell outstrips normal repair processes. Weakness and disability is progressive due to breakdown of muscle tissue. As adipose and connective tissue invades the myofibrils, pseudohypertrophy of the calf and other muscles develops (Emery, 1987). Eventually the child is dependent on a wheelchair (usually by 12 years of age) with death ensuing by late teens, although some live into their twenties with or without ventilator support.

CHAPTER 2

METHOD

Participants

Participants will be approximately 170 women over the age of 18 who, because of a family history of Duchenne muscular dystrophy (DMD), are potential carriers of the disease (an X-linked, recessive trait). The study does not include male subjects because all males with the defective dystrophin gene have the fatal illness and are not considered carriers. In the rare event of having offspring, their sons, who receive the father's Y chromosome, could not receive the affected gene, located as it is on the X chromosome. All of their daughters would be carriers.

Participants will be respondents to a questionnaire published in mailings by Parent Project for Muscular Dystrophy to their registered members. The not-for-profit organization was founded in 1994 by parents of children with Duchenne and Becker muscular dystrophy. Its mission includes funding research, disseminating information on care, state-of-the-art treatment, research findings and connecting interested parties worldwide. The estimate of 170 participants is based on a 10% response rate to mailings to the 1700 member families of Parent Project MD.

Materials

Materials will include a request to the Institutional Review Board of the University of Nevada Las Vegas for approval of the study. The packet mailed to families

registered with Parent Project MD will contain a cover letter from Parent Project MD explaining that they view the research as valuable and therefore have volunteered to offer the opportunity to participate to all members, while safeguarding the confidentiality of the mailing list. Other enclosures will include an informed consent document, the survey instrument itself (see Appendix I), and a business reply envelope or stamped return envelope addressed to the researcher.

The survey instrument items deal with feelings of stigma, the proposed mediator (perceived desirability as a mate and as a parent, present or potential), and the proposed moderators (input from family of origin, number of roles, importance placed on biological parenting, genetic testing, genetic counseling, and follow-up counseling). Most responses will require circling a number from 0 to 6 to indicate how true the participant feels the statement is of her, or simply how true a statement is in general.

Feelings of stigma. Survey statements dealing with stigmatization were selected and adapted from the HIV Stigma Scale (Berger et al., 2001) and from stigma-related questions posed to persons with epilepsy (Mittan, 1986; Westbrook, Bauman, & Shinnar, 1992). Items in this category include (1) My friends know that I am, or may be, a carrier, (2) I keep my carrier status a secret from others, (8) Generally, telling people that I am (might be) a carrier has been a mistake, (10) I talk easily to people about my carrier status, (11) Being a (potential) carrier puts me at a disadvantage in finding a mate, (18) I have been told, "You shouldn't have children."

Desirability as mate and parent. The hypothesized mechanism through which carriers experience stigmatization (either from self or others) is a diminished sense of self-worth as a prospective mate or parent. In order to tap such feelings, the following

statements will be used: (3) I feel (felt) I must tell a prospective mate about my carrier potential, (9) Being a (potential) carrier makes me unworthy to reproduce, (20) Being a (potential) carrier makes me less desirable as a mate, and (23) I feel/felt I should tell a potential mate about my carrier risk (7) before we go out, (6) when an exclusive dating relationship develops, (5) before living together, (4) before getting engaged, (3) before marriage, (2) after marriage, (1) when pregnant, (0) not at all.

Input from family of origin. Participants will indicate their level of agreement with (6) Because of DMD, other people have tried to keep me from having babies, (21) If in a relationship, My mate wants me to have a/another baby, and be asked (36) Who (by relationship—for example, cousin, husband, etc.) has been the most influential in your decision-making process with regard to bearing children?

Cautionary messages. A chart, Item 37, will help clarify and account for the majority of sources and directions of influence on the women's decisions related to reproduction. Participants will be instructed to check all 'messages' that apply for each category of person

Importance placed on biological parenting. Statements designed to indicate the participant's orientation toward biological parenting as their desired form of parenting include (5) I would like to become pregnant and have a baby, (12) Bearing children is an important part of a meaningful life for a woman, (13) When I think of being a parent, I think of rearing children, not bearing them, (16) Experiencing pregnancy is very important to me, and (17) It would be sad to die without having any children to take a little bit of myself into the future.

Role accumulation. A checklist (Item 24) yielding a numerical score (a count of the weight given different roles by the participant) will be used to assess roles. The respondent will be asked to score all of the roles she enacts in as to current importance. In order to stimulate thought a list of possibilities will include (a) contributor to family income, (b) community volunteer, (c) mother, (d) other family member (for example, daughter, wife/partner), (e) professional (for example, teacher, performer, business owner), (f) student, (g) physically active person (sports, physical exercise), (h) political worker, (i) member of a religious organization, (j) hobbyist (crafts, sewing, art, etc.). Opportunity to add other roles will be provided.

Genetic testing and counseling. Research indicates that concern over the mere possibility of being a carrier provokes a considerable degree of distress (McConkie, 2000). In some cases the concern takes on a degree of certitude based on family folklore, inaccurate understanding of genetics, or both, before actual testing has been performed and sometimes precluding it. To determine to what degree potential carriers have pursued the technologies available to obtain the greatest amount of factual information, a combination of statements and questions will be used: (7) I have tried to determine whether I am a carrier of DMD through laboratory testing, (14) I have confidence in the accuracy of laboratory carrier testing, (25) If you have had carrier testing, how old were you when you first had a carrier test? (26) What test(s) did you use? (Examples: CPK or CK, direct gene testing, linkage deletion testing, PCR, Southern blot, DGGE, etc.), (27) According to what you were told, what were the results? (28) Who presented the results to you? (29) Was there an opportunity to discuss your reaction at the time? (30) According to the test results, what is the chance you are a carrier? (31) Have you had any

additional testing? (Provide types and your age at that time if possible), (32) Have you seen a Genetic Counselor about whether you are a carrier or not?

In addition to examining the preciseness with which the participant perceives her risk factor, answers to the questions about testing will provide a snapshot of testing utilization by potential carriers of DMD and the degree to which they are served by genetic counseling specialists.

Follow-up counseling. In most settings, those who do receive genetic counseling are limited to one or two brief sessions, necessarily focused on explication of scientific information. Therefore, for help in psychologically processing the confirmation or minimization of their carrier risk, other resources would need to be explored. More important than the particular approach or discipline of the counselor would be the client's opportunity to decrease isolation and normalize their emotional responses to their circumstances. Stigmatization would be expected to be less among those who have had opportunity to do so. A combination of statements and questions will consist of (4) I would like to discuss my feelings about having babies with a counselor of some kind, (15) I belong or have belonged a genetic support group, (19) I know people with whom I discuss my feelings about genetic risk, (33) Have you sought any professional help in dealing with your feelings about the genetic information? If yes, from whom? (Check all that apply—Typical degrees for each are in parentheses), (1) genetic counselor (MS), counselor (MA, PhD, EdS), psychologist (PhD), social worker (BSW, MSW), psychiatrist (MD), (2) clergy (pastor, minister, priest), family doctor/primary care practitioner (MD, Physician's Assist., Nurse Practitioner), or specify any other, and (35) In visits with the counselors I listed above, I was able to freely express my feelings.

CHAPTER 3

PROCEDURE

The principle investigator will send to the director of Parent Project MD, Pat Furlong, MSN, originals of the documents described in the Materials section. Her staff will make up packets of the materials and mail them to all registered families. Parent Project will fund the copying, mailing, and return postage. When a qualifying member of a family receiving a packet decides to participate, she will read the material, sign and keep the informed consent, fill out the questionnaire and mail it anonymously to the researcher in the stamped, addressed envelope provided in the packet.

When the experimenter receives a response, she will assign it a number and enter the data in a spreadsheet. No record will be made of any information (such as name of city of origin or zip code of postmark) included on the envelope other than date mailed. Participants will be instructed not to write their name or return address on the questionnaire or envelope, and not to return the signed informed consent document, but to keep it for their records.

CHAPTER 4

RESULTS

Parent Project MD reported mailing approximately 1600 surveys to their contact list. The survey was accompanied by an invitation to women to participate from the president of PPMD and by a request by the researcher to share the survey with other potential carriers among their acquaintances whether or not they have had children. Contrary to expectations, PPMD did not include a business reply envelope or stamped, addressed return envelope in the packet. One hundred nineteen women participated yielding a response rate of 7.5%. One woman had experience with Becker muscular dystrophy, rather than Duchenne, so her data were not included in the analysis. Although the initial intent was to examine the attitudes of potential carriers who had not yet had children, only 11, or 9.3%, of the participants were women who did not have at least one son with DMD. Apparently, very few recipients passed the survey on to younger female relatives who were potential carriers but had not yet had children, as invited to do. Without family assistance, such women are difficult to locate as they are not likely to be directly associated with organizations geared toward parents of sons with DMD or with genetics clinics. Therefore, a mismatch occurred between the population responding and that for whom the survey was designed.

Sample Characteristics

Because mutations can occur sporadically, either in maternal gametes or in the embryo during development, a mother of one Duchenne child does not necessarily carry the gene herself. Thus, the sample consisted of both carriers and noncarriers within the same population, providing a control group made up of women in other ways similarly affected by DMD. The participants ranged in age from 21 to 56 years ($M = 40.9$; $SD = 7.1$), with one outlier, a grandmother of 79, responding (see Table 1). Approximately 30% did not consider themselves to be carriers of DMD and in 80% of the families there had been no previous cases of the disorder.

For the majority of participants, sophisticated methods of avoiding an affected child such as *in vitro* fertilization and preimplantation genetic diagnosis of embryos were evidently not an option (see Table 2). Prenatal diagnosis and termination of either any male or of affected males was utilized to a greater extent. Not all terminations, however, were related to genetic concerns.

Construction of Measures

Measurement of stigma. To obtain a single index to represent stigma, the six items pertaining to feelings of stigma were summed and averaged. Correlations among the stigma items were generally not large (see Table 3). A Cronbach's alpha of .411 was obtained from the six items (1) "My ***friends know*** that I am, or may be, a carrier of the muscular dystrophy gene," (2) "I keep my carrier status a ***secret*** from others, (8) Generally, telling people that I am (might be) a carrier has been a ***mistake***," (10) "I ***talk*** easily to people about my carrier status," (11) "Being a (potential) carrier puts me at a

Table 1

Selected Demographic Information

Variable	Number (% of sample)	(% of responses)
Age		
Under 24	5 (4.2%)	(4.3%)
25 to 34	12 (10.1%)	(10.3%)
35 to 44	65 (54.6%)	(56%)
45 to 54	29 (24.4%)	(25%)
55 and over	5 (4.2%)	(4.3%)
Considers self to be a carrier?		
No	34 (28%)	(30.1%)
Yes	67 (56.3%)	(59.3%)
Maybe	12 (10.1%)	(10.6%)
Cases of DMD in Previous Generations of the Family?		
No	94 (79.0%)	(79.7%)
Yes	24 (20.2%)	(20.3%)
Older Brother(s)	7	
Younger Brother	10	
Uncle	2	
Cousin(s)	5	
Children with Duchenne?		
None	11 (9.2%)	(9.5%)
One	98 (83.1%)	(85.2%)
Two	7 (5.9%)	(6.1%)

Table 2

Utilization of Reproductive Technologies

Variable	Number (% of sample)	(% of responses)
Contraception		
None	30 (25.4%)	(25.9%)
Temporary	38 (32.2%)	(32.8%)
Permanent	48 (40.7%)	(41.4%)
Prenatal Diagnosis	28 (23.7%)	(25.2%)
Termination	22 (25.4%)	(20%)
in vitro Fertilization	1 (0.8%)	(0.9%)
Natural Sex Selection	7 (5.9%)	(6.1%)

Table 3

Intercorrelations Between Stigma Items

Items	Friends	Secret	Mistake	Talk easily	Disadvantage	Shouldn't
			(n = 91)			
Friends	—	–.13	–.04	.30	.15	.13
Secret		—	.29	–.57	.02	.15
Mistake			—	–.24	.31	.31
Talk easily				—	–.17	.12
Disadvantage					—	.24
Shouldn't						—

disadvantage in finding a mate," and (18) "I have been told, 'You ***shouldn't*** have children.'" The strongest correlation was between ***secret*** and ***talk***, –.573.

In view of the poor correlations and alphas, a reconceptualization of the psychological impact of carrier status was in order. The construct of stigmatization, in this study at least, has proven elusive. Items meant to tap into it on the basis of a desire to keep a "spoiled identity" hidden did not do so successfully. Combining two statements that are conceptually related to stigma created an alternative stigma index: (2) ***secret*** and (8) ***mistake***. If women have found that it has been a mistake to tell others that they are carriers, they are likely to keep it a secret from that time on.

Measurement of desirability. The same process was followed for the four items regarding desirability as mate (see Table 4), (3) "I feel/felt I must ***tell*** a prospective mate about my carrier potential," (9) "Being a (potential) carrier makes me ***unworthy*** to reproduce," (20) "Being a (potential) carrier makes me ***less desirable*** as a mate," and (23) "I feel/felt I should tell a potential mate about my carrier risk…" which indicated how early in a relationship disclosure of carrier status should be made (***tell early***). The Cronbach's alpha among these items was .56.

As with the stigma index, items were regrouped to combine two statements relating to desirability as a mate (11) ***disadvantage*** and (20) ***less desirable***. Being less desirable as a mate certainly puts one at a disadvantage in finding a mate. These two items were originally not grouped together, however they have an alpha coefficient of .85 and both clearly relate to the same concept. In terms of raw responses, women who believe they carry the gene said that it was a least somewhat true (rated 3 or above) that being a carrier

made her unworthy to reproduce (40.6%), less desirable as a mate, (42.3%), or at a disadvantage in finding a mate (37.9%).

Table 4

Intercorrelations Between Desirability Items

Item	Tell mate	Unworthy	Less desirable	Tell early
		(n = 83)		
Tell mate	—	.14	.05	.28
Unworthy		—	.60	.11
Less desirable			—	.12
Tell early				—

Measurement of Moderators

Cautionary messages. The last three "messages" on the family input chart give an indication of communication from the woman's relatives who "Implied that my chances were too high to risk having children," "Recommended adopting," or "Recommended childlessness." The cautionary message scores (which also included "Recommended carrier testing" and "Arranged carrier testing") from all relatives or significant others were summed to arrive at one score (***total messages***) per participant.

Importance of biological parenting. Measuring the importance placed on biological parenting were five items: (5) "I would like to ***become preg***nant and have a baby," (12) "Bearing children is an important part of a ***meaningful*** life for a woman," (13) "When I think of being a parent, I think of ***rear***ing children, not ***bear***ing them," (16)

"Experiencing pregnancy is very ***important to me***," and (17) "It would be sad to die without having any children to take my family ***bloodline*** into the future" (see Table 5). A Cronbach's alpha of .54 was obtained. Item 5 was removed because wanting to become pregnant is affected by many personal situational details, e.g. age, size of family, marital status, whereas ideally the biological parenting index should measure general attitudes rather than specific intentions. Item 13 did not indicate a preference for biological parenting, perhaps because women might feel child rearing is more central to parenting than child bearing even though pregnancy is very important to them. When items 5 and 13, with correlations to other items under .40, were removed, the coefficient alpha improved to .71.

Table 5

Intercorrelations Between Biological Parenting Items

Item	become preg	meaningful	rear vs. bear	imprt to me	bloodline
		(n = 103)			
become preg	—	.11	.02	.18	.11
meaningful		—	.11	.52	.41
rear vs. bear			—	.01	.03
important to me				—	.46
bloodline					—

Mediational Analysis

It was hypothesized that the relationship between carrier status and stigmatization would be mediated by desirability as a mate. To maintain a clear distinction between

groups, participants who answered "not sure" or "maybe" in response to "Do you consider yourself to be a carrier of Duchenne muscular dystrophy?" were eliminated from analysis.[1] As the first step in Baron and Kenny's (1986) moderated mediational analysis procedure, the variable Carrier was regressed on the reformulated Stigma index (see Table 6). Then both Carrier and the Desirability variable were regressed on Stigma.

Table 6

Summary of Mediational Regression Analysis for Variables Predicting Stigma (N = 80)

Variable	Standard Coefficient	*t*	*p*	R^2
Carrier status	0.234	2.130	.04	.055
Carrier status	0.193	1.795	.08	
Desirability	0.273	2.536	.01	.128

There was no significant reduction in the standard coefficient (.234 was reduced to .193), indicating that perceived desirability as a mate was not mediating the relationship between carrier status and stigma. No mediational effect having been found, no further steps in the Baron and Kenny procedure were carried out.

Moderational Analysis

A further hypothesis was that the reception of cautionary messages from family and others closely involved would moderate the impact of carrier status on stigma.

[1] A data set was created from those who answered "yes" or "no" regarding carrier status and also answered items 2, 8, 11, and 20, n = 78.

Specifically, stronger cautionary input from more sources would increase the sense of stigma. The stigma index was used to examine the moderational effect of cautionary messages from family and other significant persons. In order to test this, a moderated multiple regression analysis was performed in which stigma was regressed on carrier status, cautionary messages, and the interaction term of carrier status and cautionary messages (see Table 7). Analyzing carriers and noncarriers, a significant interaction was found to exist between carrier status and cautionary messages ($p = .035$). Such messages were not predicting stigma among noncarriers as they were doing among carriers. Consistent with the hypothesis, there was a significant effect for carrier status (see Figure 1).

Table 7

Summary of Moderated Multiple Regression Analysis (N = 91)

Variable	*B*	*t*	*p*	*R2*
Carrier	.008	.016	.99	
Messages	-0.134	-1.15	.25	
Carrier X Messages	.271	2.15	.04	.121

The same moderated multiple regression analysis was used to examine other proposed moderaters. No significant effects were found.

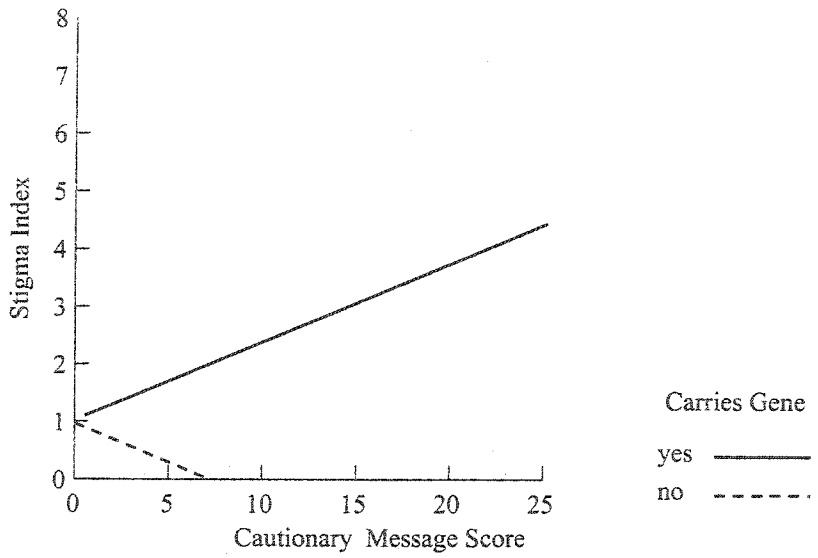

Figure 1. Cautionary messages predict stigma for carriers but not for noncarriers.

CHAPTER 5

DISCUSSION

Little support for the mediational hypotheses was found. Neither feelings of unworthiness to reproduce nor undesirability as a mate mediated feelings of stigma among the sample population as a whole. The moderational hypotheses fared only slightly better. Although no support was found for a moderating effect of the salience of biological parenting, role accumulation, genetic testing, or follow-up counseling, input from family (cautionary messages) did moderate the impact of being a carrier on stigma as measured by disclosure management.

The significant moderational effect cautionary messages had upon the carrier women's reported sense of stigma suggests that such input heightens a woman's consciousness of the negative societal evaluation of carrying the deleterious gene and thus increases her awareness of possible social disqualification and limited opportunities in regard to marriage and childbearing. Realizing that one's social identity has suffered a negative change is the essence of stigma (Berger et al., 2001).

Explanation for the lack of support for the majority of hypotheses may be found in limitations of the sample, the wording of items, the construct of stigma, or a combination of the three. The central limitation to the study stems from problems with the sample. The survey was designed primarily for women who were dealing with concerns about carrier status *prior to* bearing children. Such a sample would allow taking

a close look at prospective reproductive decision making and at what being a carrier (or the possibility of being a carrier) means for a woman's view of her desirability as a partner. It was in this context that stigmatization was conceptualized as deviance with regard to societal expectations that a woman will bear children and that the children will conform to standards of normality. The majority of carriers responding, however, were women who discovered their carrier status only due to the birth of a child with Duchenne. Thus, although dealing with the question of whether additional children could also be affected, their concern was not situated during the life phase of mate-selection when possessing a faulty gene could be most damaging to successful negotiation of a developmental stage (Erikson, 1963).

The fact that the sample population actually obtained was not the population targeted meant that many items were inappropriate for the majority of participants. Married or cohabiting women (89% of the sample) had no way of knowing, for instance, whether they should respond to items such as "Being a (potential) carrier puts me at a disadvantage in finding a mate" in hypothetical terms or, indeed, at all. The situation resulted in missing data as well as misleading responses. Difficulties in interpretation ensue. Rating "I would like to become pregnant and have a baby" as "0—not true" on the Likert-type scale would signify something entirely different in the case of a woman who had finished family-building than for a younger woman who had not had any children. A number of the respondents realized this, as did the woman who commented, "I have not dealt with most of these issues. I have been married for 20 years and had no history of DMD in my family until my son was diagnosed 5 yrs. ago."

Another complication was the different perspective from which noncarriers would consider the items than that from which carriers would answer them. Women were invited to participate if they were among those who *could* be carriers based upon their relationship to a Duchenne patient.[2] Thirty percent of the women answering the question "Do you consider yourself a carrier of DMD?" replied "no," 11% responded "maybe" or "not sure," while 59% said "yes." Because not all respondents were carriers and of self-identified carriers, only four had not had children, the target population was not reached. Items meant to access feelings of stigmatization based on disclosure management, for instance, were confounded. If a woman's carrier status were negative, she would likely not keep that status secret from others. It is similarly unlikely that she would have found telling others to have been a mistake. One participant commented, "Most questions assume carrier status is positive or unknown. I hope my answers are not misleading because I believe that I am not a carrier." Although the noncarrier subjects served importantly as a control group, the number of experimental subjects was therefore reduced to 30% fewer than the total number of respondents.

The items could have been better worded to allow for the fact that a proportion of the respondents did not consider themselves carriers. Item 3, "I feel (felt) that I must tell a prospective mate about my carrier potential," allowed for a retrospective report providing that the woman knew that she was a carrier before marriage. In the sample, however, such was not typically the case. In addition, however, if she were not a carrier, "my carrier potential" would be a non-issue that she might tell her beau about, but doing so

[2] Geneticist's traditional rough estimate is that one-third of patients with X-linked disorders come from a family with the trait, one-third are the result of *de novo* mutations in a male, resulting in a new case, and one-third are the result of *de novo* mutations in a female, resulting in a new carrier who becomes the mother of the Duchenne patient.

would not carry the same implication that it would for carriers. Among the latter, Kay and Kingston (2002) found that the women they interviewed felt duty-bound to give partners "a chance to get out of it if they were scared (p.175)." Such anticipation of negative reactions in response to a deviance from normality is an integral characteristic of stigma (Berger et al., 2001), but the variation in responses possible from already married women and from noncarriers may have obscured correlations in the data.

Aside from the limitations due to the makeup of the sample population, the items themselves presented difficulties. Low means and a lack of variability on many items seriously limit the potential for significant differences. For instance, among definite carriers the mean on Item 2 (***secret***) was 0.894 with a standard deviation of 1.637, on Item 8 (***mistake***) 0.697 and 1.358, on Item 15 (***support***) 0.881 and 1.847.

The fact that few women reported appreciable agreement with statements indicative of stigma does, however, address the subject of interest. It is apparent that a strong sense of being stigmatized is not characteristic of carriers in general. However, some participants did express themselves in the comment section along the lines of this observation: "I do feel some level of shame and some sense of punishment for having both of my sons with DMD. I do think I blame myself, but there is no family history of DMD so there was no way I could have known. I chose not to have any other children, but I did grieve this decision."

Furthermore, the difficulty in measuring stigmatization may also be due to the vagueness of the construct itself. Disentangling feelings of negative self-evaluation from the recognition that one's characteristics may result in negative evaluation by others presents a difficult challenge. The statements "Being a (potential) carrier makes me

unworthy to reproduce" and "Being a (potential) carrier makes me less desirable as a mate" were the most highly correlated of those in the original stigma grouping. Although the first refers to self-evaluation and the second to how potential mates might evaluate one, worthiness may be closely connected with the opinions of others and, hence, perceived stigma. The model of perceived stigma (Berger et al., 2001) proposes that perception of negative societal attitudes toward people with the trait in question sets the stage; then perception of stigma results from "awareness of actual or potential social disqualification, limited opportunities, and negative change in social identity (p. 520)." Knowing that negative societal attitudes exist is thus distinguished from recognizing the effect they may have of limiting social interaction and spoiling social identity. Measuring perceived stigma would reasonably encompass items that were used in the stigma, desirability as a mate, and worthiness to reproduce categories in the present study. In sum, it may be that stigma as a construct is not different enough from negative self-evaluation and societal discrimination to function independently. In this study, the decision was made to consider attempts to hide the fact of being a carrier as indication that the possessor was aware of the negative social impact the mark could have and managed disclosure due to a sense of stigmatization (Goffman, 1963).

Inadequate Testing

A collateral finding of interest that emerged from the data was that determination of carrier status for DMD was far from straightforward for the participants. A number of women had been tested only by measuring their creatine (phospho)kinase (CK or CPK) level (see Table 8).

Table 8

Tests Used to Determine Carrier Status

Type of Test	Number (% of sample)	(% of responses)
Don't remember	9 (7.6%)	(9.5%)
CK	29 (24.4%)	(30%)
DNA (all types)	57 (47.9%)	(60%)

Creatine kinase is an enzyme that leaks out of muscle cells lacking dystrophin resulting in unusually high blood levels of the enzyme. However, "...CK test utility is limited to those suspected carriers whose results fall above the healthy population interval. A low CK result does not provide sufficient assurance of noncarrier status" (Gruemer et al., 1985). In addition, false positives can result due to minor increases in CK from physical exercise. In approximately 70% of carrier women CK levels are elevated, but in roughly 30% the level will be within normal range (C. Strickland, personal communication, March 20, 2004).

Some of the women surveyed had become pregnant thinking that they were not carriers on the basis of CK testing when in fact they were; some refrained from having children thinking they were carriers, when in reality they may not be. Some respondents are considering tubal ligation solely on the basis of CK testing. Checking levels of creatine kinase does not constitute a *genetic* test as doing so does not examine the dystrophin gene.

DNA testing takes two forms. Linkage analysis relies on knowledge of a number of family members' "markers" associated with a dystrophin mutation. If a woman's X chromosome contains those markers, chances are high that she carries the genetic

mutation. Linkage analysis is done when there is more than one affected individual in the family and the actual DNA mutation causing the disease is unknown. A second type of test can be run by direct analysis of the chromosome, either through Quantitative PCR or through checking the base pair sequences against the sequences found in the normal dystrophin gene. If the patient's mutation is known, matching alterations in the relative's gene provide definitive determination of carrier status. Without information regarding the precise mutation in the related Duchenne patient, errors in sequencing may masquerade as mutations, resulting in false positives.

Testing among women affiliated with PPMD may be among the best testing experienced by persons affected by DMD. Effective advocacy groups must have social capital, thus tend to be composed of educated persons with reasonable income and status (Epstein, 1995). The fact that even among this group of informed, proactive, relatively sophisticated persons decisions are being made on the basis of clearly inferior sources of information (CK testing over DNA) is cause for concern.

Rate of Spontaneous Mutation

Among participants, 88% of mothers of Duchenne patients reported that there had not been any occurrence of the disorder in previous generations. The traditional approximation has been that two-thirds of Duchenne cases occur in families with no previous history. Thus, the rate of spontaneous mutation may have been underestimated. On the other hand, the number of affected children born to women positive for family history may have decreased, perhaps through awareness of genetic risk. Another possibility is that women without a family history of the disorder are overrepresented in the sample. It may be that women who are acquainted with the disorder through family

experience are less optimistic that research will find a cure, or more knowledgeable about what to expect and services available, thus not as moved to join informational and advocacy groups. Epidemiological studies, only recently begun by the Center for Disease Control and Prevention (National Center for Birth Defects and Developmental Disabilities, ¶ 3) are necessary before conclusions about the discrepancy are warranted.

Marital Stability

Another concomitant finding of interest involves spousal relationships. Marital stability has traditionally been thought to be problematic among parents of DMD children, with a high divorce rate postulated. Among participants, however, three out of four of the mothers of boys with Duchenne are still married to their first spouse. The divorce rate (participants whose current marital status is *divorced* divided by total participants) of the sample (including ages 25 to 54) is 1,611 per 100,000 (age-adjusted). The same cohort's rate from the 2002 U.S. census data is 5,936 per 100,000. The participants, therefore, have a divorce rate that is .27 of the national average among women 25 through 54. Interpretation of the low divorce rate must include the particular nature of the sample which limits other generalizations of the study. Except in cases where the husband reacts with bitterness toward the wife or with denial and rejection of the extremely stressful situation, it may be that heightened concern for the child they love in common protects the marriage.

CHAPTER 6

CONCLUSION

The study was undertaken to extend previous research indicating that carriers of genetic disorders do not exhibit lowered self-esteem (Denayer et al., 1996), with the possible exception of in the limited area of "future parent role" (McConkie, 2000). It was assumed that in the case of X-linked disorders, as opposed to autosomal recessive traits, the undiffused genetic "responsibility" for the disability would promote a more discernable sense of stigmatization in the limited area of desirability as a partner and value in the marriage marketplace. While feelings of stigmatization are far from prominent in the sample, the findings do reveal that in a fine-grained examination of carrier women, the incidence of cautionary messages from family and others does in fact moderate the carriers' evaluation of their desirability as a spouse. The more relatives and other closely involved persons suggest that her risks preclude future childbearing, the more a carrier will feel devalued as a worthy partner. Whether a sense of being less desirable and at a disadvantage in finding a mate is conceptualized as stigmatization or as the result of societal discrimination (if indeed there is a difference between the two concepts), the absence of moderating effects from other factors highlights the relative importance of supportive communication between family members (and professional contacts). Expressions that preempt the childbearing prerogatives of the women involved appear to be connected with diminished sense of desirability as a mate. Although the data

did not specify the age at which such messages were received, research in adolescent psychology emphasizes the importance to identity development in the teen years of feeling that one has the prerogative to make one's own choices (Zastrow & Kirst-Ashman, 1990, chap. 6).

In future research, the influence of ethnocultural background should be investigated. The limits of acceptable female roles vary widely, with the importance of childbearing being more pronounced in agrarian, developing societies than seems to be the case in many industrialized, modern nations. A Pakistani-British woman whose brother has DMD stated,

> "In our society it was viewed as a disease, as... you know, you know, don't go near her! You know she's not...[she's] spoiled, not good! Backward people, obviously people with no knowledge in genetics, or science, or anything, they assumed that I was a bad, bad apple, basically. You know, so yes, it did worry my mother and worry...Oh my god, how am I going to get her married off? ... Cause, first impression, you look at him and think, ooo, you know, don't, they don't treat you normal. No matter what people say they don't.
>
> Q. So it isn't a question of 'Oh, is this genetic, is this hereditary? And are you a carrier?' It's just that it's in the family?
>
> A. Oh, it's stamped! Yeah. Oh, especially if they find out it's in the family!
>
> Q. Uh huh. But even when it's not known for sure...
>
> A. Oh, in our culture..
>
> Q. It's just sort of assumed?
>
> A. Oh definitely assumed that um 'No. All her kids will be bad' (Karwoski, 2003)."

The study indicates that cautionary messages communicated to women who carry the muscular dystrophy mutation moderate the degree of stigma they experience.

Societies in which there is great emphasis placed on women's role as bearers would likely produce greater expression of caution and need for concealment, hence a greater level of stigmatization. Application of lessons learned regarding the conceptualization and measurement of stigma in this study may contribute to more definitive results in further research efforts, particularly in the context of multicultural comparisons.

APPENDIX I

RESEARCH MATERIALS

DESCRIPTION OF STUDY

Name: Jane Karwoski
Department: Psychology
Title of Study: The Stigma of Carrier Status [referred to as Genetic Risk and Self-Concept on documents for the participants]

1. **SUBJECTS:**

Participants will be approximately 170 women over the age of 18 who, because of a family history of Duchenne muscular dystrophy (DMD), are potential carriers of the disease (an X-linked, recessive trait). The study does not include male subjects because all males with the defective dystrophin gene have the fatal illness and are not considered carriers. In the rare event of having offspring, their sons, who receive the father's Y chromosome, could not receive the affected gene, located as it is on the X chromosome. All of their daughters would be carriers.

Participants will be respondents to a questionnaire published in mailings by Parent Project for Muscular Dystrophy to their registered members. The estimate of 170 participants is based on a 10% response rate to mailings to the 1700 member families of Parent Project MD.

2. **PURPOSE, METHODS, PROCEDURES:** Describe in detail the purpose, research methods, and procedures of the study.

Purpose: As genetic testing becomes available for more and more diseases it is crucial that we learn more about the psychological impact of carrier status determination. The availability and comprehensiveness of genetic counseling vary considerably. A major purpose of the present study is to assess the degree to which potential carriers of one hereditary disease, Duchenne muscular dystrophy, experience stigmatization due to being at risk of passing on the disease and the impact on self-concept as a parent or potential parent. The study will also measure the degree and source of outside influence on childbearing decisions. A subsidiary goal is to assess to what degree potential carriers of DMD receive genetic testing and counseling.

It is hypothesized that a greater sense stigmatization will be evident in the self-concept of women for whom biological parenthood is of paramount importance, who have a low level of self-complexity, whose decisions have been usurped by family members or others, and who have received little or no counseling support.

This study offers a rare opportunity for potential carriers to tell their story, albeit in an abbreviated manner. That relatives do appreciate interest in their experience (when normally all attention is focused on the difficulties suffered by the patient) is borne out by responses to similar studies relating to cystic fibrosis (CF) by Joanna Fanos, PhD, UCSF and fragile X by Allyn McConkie-Rosell, PhD, Duke University Medical Center.

Pending further developments in gene therapy to counteract biological deficiencies resulting from defective genes, prevention, in the form of avoiding births of affected offspring, is the only way to reduce the individual and societal costs of hereditary disease. Problems associated with such a prevention approach include accuracy rate in identification of carriers, prenatal diagnosis accuracy, risks to mother and fetus, and psychological adjustment to carrier status. This study will address the neglected area of the psychosocial adjustment to carrier status.

Method: A survey instrument will be used to collect information regarding the experience and attitudes of women at risk of being DMD carriers. Responses will be tabulated and analyzed using SPSS. Correlations between responses to stigma items and items tapping family influence, self-complexity, possible parental role, amount and type of testing and counseling are of particular interest. Structural Equation Modeling will be employed in further interpreting the data.

Procedure: The attached cover letters (from Parent Project MD and from the researcher), informed consent document, and anonymous questionnaire will be reproduced and mailed by Parent Project for Muscular Dystrophy, Middletown, Ohio, to their registered members along with a returned envelope stamped and addressed to the researcher. Those recipients who meet the eligibility criteria and elect to participate will complete the informed consent document and save it for their own records. They will indicate their response to all questions they choose to answer and return it to the researcher who will identify the returned questionnaires by number for the purpose of data analysis. Additional packets will be available upon request from Parent Project MD for the duration of the study.

3. **RISKS:** There will be minimal risk to the participants in the study. The risks associated with the study are less than what an individual would normally be exposed to in discussing the same subject in private, given the anonymity of the responses. If the issues dealt with by the survey are emotionally distressing to an individual, the distress is likely pre-existing and a request to share feelings in an anonymous setting may present a welcome opportunity for self-expression. Surveys will be completed in private by the participant at her own pace, then returned to the investigator by mail without identifying information.

Data files will not include name, address, or any other identifying information associated with the participant and will be stored on a secure disk. Each data file will be coded with an identification number for the participant based upon order of receipt. There will be no

lab records associating ID number and the respondent's name or other personally identifying information.

4. **BENEFITS:** The primary direct benefit to participants will include a basic understanding of the purpose of the study, and an appreciation for the general research area. The participants also will be provided with contact information for obtaining a summary of average (NOT individual) results from the study, if desired. The self-exploration inherent in responding to the survey and the opportunity to contribute to research aimed at lessening the psychological distress associated with DMD may contribute to the participants' adjustment to familial disease. Content of the questionnaire itself will aid respondents to be better informed regarding benefits of genetic counseling and possible courses of action of which they may have been unaware.

Additional benefits will accrue to the social psychological study of self and identity. For the growing number of genetic counselors, the study should shed light on what increases or decreases a sense of stigmatization among their clients.

The identification of genetic risk has stimulated international debate regarding the advisability of large-scale screening. It is therefore crucial to assess and address the related potential for, and mechanisms of, stigmatization processes.

5. **RISK-BENEFIT RATIO:** Since participation in the study is voluntary and is determined with foreknowledge of the questions asked, those for whom participation may be problematic will presumably not participate. Participants will gain the benefit of seeing an interest taken in their needs and knowing that their own contribution could conceivable lead to improvement in services. Therefore benefits far outweigh risks.

6. **COSTS TO SUBJECTS:** The only clearly identifiable cost to each participant will be in terms of the required time commitment of a single 30-minute session for reading the cover letters, consent document and completing the survey. No funding is available to financially compensate participants.

7. **INFORMED CONSENT:** Informed consent will be obtained directly from each participant. Each potential participant will be required by the experimenter to carefully read a statement of informed consent (see attached form). The experimenter will be the principal investigator.

The attached statement of informed consent provides participants with a broad description of the kind of task they will be asked to perform and will be mailed with the questionnaire. In adhering to APA ethical guidelines, the informed consent form also reminds potential participants that they can withdraw from the experiment at any time. The investigator's E-mail address and telephone number are noted on the form with an invitation to contact her with questions about participation. After any questions have been addressed, those choosing to participate will sign and date a copy of the form for their personal records. (Anonymity of participants is assured both during and after this 3-year period due to the safeguards previously described.)

8. **CHILD/YOUTH ASSENT:** No one under 18 years of age is eligible to participate.

Cover letter from Parent Project Muscular Dystrophy

August 15, 2003

Pat Furlong, President
Parent Project Muscular Dystrophy
1012 North University Boulevard
Middletown, Ohio 45042

Dear Friends,

This mailing contains a research questionnaire focused on the impact of DMD on women. Although some of the questions may be difficult, we feel that your answers will be helpful in understanding the effect of the disease on women and their self-concept regarding reproductive decisions. The concern and uncertainty surrounding carrier status is a largely unacknowledged source of distress and strain on families with DMD. We need to know more about how risk is perceived and communicated within families and between women and healthcare providers. Findings may help parents know how best to help their daughters, as well as in considering more children of their own.

Jane and I have been communicating about her research for almost a year now. She has dealt with these issues herself and will treat your views with the utmost respect. The response is entirely anonymous, so your confidentiality is assured. The survey materials can be photocopied for any other women over 18 years of age who are at risk of passing on DMD and wish to participate. Please return the completed questionnaire directly to Jane as soon as possible. Although all responses will be helpful, you may omit questions you do not wish to answer. Do not put a return address on the envelope. Results will be summarized in the future on the Parent Project MD website.

Sincerely,

Patricia Furlong, President

Cover letter from researcher

August 15, 2003

Dear Friends,

My name is Jane Karwoski. I'd like to have your help with something of interest to all of us. My brother, Alan, was diagnosed with DMD when he was four years old and I was just two. Everything that happened after that affected me as well and changed my life forever. I have arrived at a point in my life where I realize that I might use my experience, coupled with my education and reflection over the years, to look for ways to assist other potential carriers of DMD.

I am interested in how concern about passing on DMD to one's children affects the way we think and feel about ourselves and how it affects our decisions about being parents. For many, being a carrier was discovered only with the birth of an affected child. For others, like me, before ever having children we became aware that we may face a risk, due to a relative having the disorder. The distress of DMD includes that of family members for whom the natural anticipation of having a family gets very complicated indeed. When we consider the lives of children yet to come, we hope they will not have genetic disorders. How do we manage that concern? How does being a carrier, or possibly being a carrier, affect one's self-concept and decisions?

The answers are important because they may reveal whether women are receiving appropriate support from health professionals and may point to how they can be better served in dealing with the impact of hereditary disorder. The answers may also help parents understand how to best help their daughters deal with the possibility of being carriers.

My survey will try to get at several issues that I suspect may be involved. For this study I intend to collect responses by means of a survey mailed to families associated with Parent Project MD. Once I have collected sufficient responses over a month or two, I will analyze the data and make the findings known through mailings or the parentprojectmd.org website. Your privacy will be respected at all times. No responses will be identified with any individual participant. The study will be the basis of my master's thesis, and potentially an article or two in research journals devoted to genetic counseling. It is important for genetic counselors to understand how a person feels about the chance of being a carrier of DMD and the impact that has on her life.

I hope to expand the study for my dissertation, incorporating improvements that your responses help me make. Since most of you visit the website because you have a family member with DMD or are diagnosed with DMD, I'd like to ask your help not only through participation, but through informing other adult female relatives (sisters, aunts, cousins) who might not be in touch with Parent Project MD. Women of any age over 18 are encouraged to participate whether or not they have had children or know for sure that

they are a carrier. More details regarding eligibility and informed consent appear with the survey included with this mailing.

Sincerely,

Jane Karwoski, MSW
PhD student
Experimental (Social) Psychology
University of Nevada, Las Vegas

University of Nevada, Las Vegas

Department of Psychology

INFORMED CONSENT

General Information:
I am Jane Karwoski from the UNLV Department of Psychology. I am the researcher on this project. You are invited to participate in a research study. The study is about how the possibility of passing Duchenne muscular dystrophy (DMD) on to her children affects a woman's view of herself as a woman, parent, or potential parent. Women who *may* be carriers include the mother, sister, aunt, grandmother, niece, or cousin, of a person (usually male) that has been diagnosed with DMD. Please note that because of the way the DMD gene is passed on, not all such relatives are carriers. But they are all invited to participate. You do NOT need to have children to take part. Whether you have no children, do have children (but none with DMD), or have children with Duchenne muscular dystrophy, your views will be appreciated.

Procedure:
If you volunteer to participate in this study, you will be asked to do the following:

Fill out an anonymous questionnaire, providing information on

- your background in relation to DMD
- carrier testing you have or have not had
- your feelings about parenthood.

Please examine the questionnaire to gain a complete understanding of what information will be requested, keeping in mind that you are not obligated to answer any question you do not want to answer.

Benefits of Participation:

By participating you will have the opportunity to share your perspective on how DMD has affected family members of Duchenne patients (female relatives such as yourself). You may also receive an increased understanding of options available to you such as genetic risk assessment, counseling options, and ongoing research that aims to help women make their own reproductive decisions.

Risks of Participation:

You may experience some degree of emotional distress if the issues involved have been difficult for you. You might be uncomfortable answering some of the questions asked. You are welcome to discuss this with me. I will explain the questions to you in more detail. Although answers to all questions will be helpful to the researcher, you may omit any particular questions if you wish.

Contact Information:

If you have any questions about the study or if you experience harmful effects as a result of participation in this study, you may contact me by phone at 702-597-3313 or by E-mail at jkar_unlv@hotmail.com or by mail at P.O. Box 72544, Las Vegas, NV, 89170-2544.

For questions regarding the rights of research subjects, you may contact **the UNLV Office for the Protection of Research Subjects at 702-895-2794.**

Voluntary Participation:

Your participation in this study is voluntary. You may refuse to participate in this study or in any part of this study. You may withdraw at any time without prejudice to your relations with the university. You are encouraged to ask questions about this study at the beginning or any time during the research study.

Confidentiality:

In order to maintain anonymity, please do not put your name or address on the questionnaire or the return envelope. No reference will be made in written or oral materials that could link you to this study. All records will be stored in a locked facility for at least 3 years after completion of the study, until they are destroyed.

Participant Consent:

I have read the above information and agree to participate in this study. I am at least 18 years of age. I will keep this form for my own records.

_____ _____

Signature of Participant Date

Jane Karwoski, MSW c/o Parent Project MD, 1012 North University Blvd, Middletown, OH 45042

For the following items, please circle the number between 0 and 6 below each statement to indicate how true you think the statement is of you. For example, circling "0" means you don't think it is true of you at all, or that in your opinion the statement is not at all true. If not completely true, but almost, you could circle "5." Right in the middle is "3." Less true would be "2" or "1." Slightly more true of you than just somewhat would be "4," and so on.

1. My friends know that I am, or may be, a carrier of the muscular dystrophy gene.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

2. I keep my carrier status a secret from others.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

3. I feel (felt) I must tell a prospective mate about my carrier potential.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

4. I would like to discuss my feelings about having babies with a counselor of some kind.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

5. I would like to become pregnant and have a baby.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

6. Because of DMD, other people have tried to keep me from having babies.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

7. I have tried to find out whether I am a carrier of DMD through laboratory testing.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

8. Generally, telling people that I am (might be) a carrier has been a mistake.

0	1	2	3	4	5	6
not true			somewhat true			very true

9. Being a (potential) carrier makes me unworthy to reproduce.

0	1	2	3	4	5	6
not true			somewhat true			very true

10. I talk easily to people about my carrier status.

0	1	2	3	4	5	6
not true			somewhat true			very true

11. Being a (potential) carrier puts me at a disadvantage in finding a mate.

0	1	2	3	4	5	6
not true			somewhat true			very true

12. Bearing children is an important part of a meaningful life for a woman.

0	1	2	3	4	5	6
not true			somewhat true			very true

13. When I think of being a parent, I think of rearing children, not bearing them.

0	1	2	3	4	5	6
not true			somewhat true			very true

14. I have confidence in the accuracy of carrier testing.

0	1	2	3	4	5	6
not true			somewhat true			very true

15. I belong or have belonged to a genetic support group.

0	1	2	3	4	5	6
not true			somewhat true			very true

16. Experiencing pregnancy is very important to me.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

17. It would be sad to die without having any children to take my family blood line into the future.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

18. I have been told "You shouldn't have children."

0	1	2	3	4	5	6
not true			somewhat true			very true

---

19. I know people with whom I can discuss my feelings about genetic risk.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

20. Being a (potential) carrier makes me less desirable as a mate.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

21. If in a relationship: My mate wants me to have a/another baby.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

22. I am satisfied with my decisions about pregnancy so far.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

23. I feel/felt I should tell a potential mate about my carrier risk
___before we go out
___when a steady or exclusive dating relationship develops
___before living together
___before getting engaged
___before marriage
___after marriage or living together
___when pregnant
___not at all

---

24. Rate the following roles as to how important you feel they are in your life at present. Add any others that matter, but are not on the list:

	Not at all			Somewhat			Very Important
contributor to family income	0	1	2	3	4	5	6
community volunteer	0	1	2	3	4	5	6
mother	0	1	2	3	4	5	6
family member (daughter, wife/partner)	0	1	2	3	4	5	6
professional (teacher, performer, business)	0	1	2	3	4	5	6
student	0	1	2	3	4	5	6
physically active person (sports, exercise)	0	1	2	3	4	5	6
political worker	0	1	2	3	4	5	6
member of a religious organization	0	1	2	3	4	5	6
hobbyist (crafts, sewing, art, etc.)	0	1	2	3	4	5	6
other: _____	0	1	2	3	4	5	6
other: _____	0	1	2	3	4	5	6
other: _____	0	1	2	3	4	5	6

If you have had carrier testing:

25. How old were you when you first had a carrier test?

26. What test(s) did you use? (Examples: CPK or CK, direct gene testing, linkage deletion testing, PCR, Southern blot, DGGE, etc.)

27. According to what were you told, what were the results?

28. Who presented the results to you?

29. Was there an opportunity to discuss your reaction at that time?

30. According to the test results, what is the chance you are a carrier?

31. Have you had any additional testing? (provide types and your age at that time if possible)

32. Have you seen a Genetic Counselor about whether you are a carrier or not? No Yes

33. Have you sought any professional help in dealing with your feelings about the genetic information?

No
Yes: From whom? (Check all that apply.) (Typical degrees for each are in parentheses.)

34. ___ genetic counselor (MS)
___ counselor (MA, PhD, EdS)
___ psychologist (PhD)
___ social worker (BSW, MSW)
___ psychiatrist (MD)

___ other ___ clergy (pastor, minister, priest)
___ family doctor/primary care practioner (MD, Physician's Assist., Nurse Practitioner)
___ specify any other:_____

---

35. In visits with the counselors I listed above, I was able to freely express my feelings.

0	1	2	3	4	5	6
not true			somewhat true			very true

---

36. Who (by relationship—for example, cousin, husband, etc.) has been the most influential in your decision-making process with regard to bearing children?

Please fill in the chart to show what family members communicated to you about pregnancy. There is room below the chart to clarify relationships or attitudes if you wish.

37. Check all 'messages' that apply for each category of person

Message about having children of my own:	Mother	Father	Aunt/Uncle	Grandparent	Other	Nonrelative
Never mentioned it						
Implied that my risk was very low or non-existant and I need not consider it						
Recommended carrier testing						
Arranged carrier testing						
Indicated that reproductive decisions were entirely up to me						
Implied that my chances were too high to risk having children						
Recommended adopting						
Recommended childlessness						
Other:						

Demographics

38. Do you consider yourself to be a carrier of Duchenne muscular dystrophy? No Yes

39. What is your date of birth? (mo/day/year) ____/____/____

40. Who in your family has/had Duchenne muscular dystrophy? (Write in the how many of each)
___Older Brother(s) ___Younger Brother(s) ___Son(s) ___Uncle(s) ___Cousin(s)
___ Grandson(s) ___Other(s):

41. Are you ___Single
___Married
___Living with other long-term partner

42. Have you been divorced? No
Yes: How many times? ____

Reproductive history

43. Have you ever been pregnant? No
Yes: How many times?_____

44. Have you borne children? No
Yes: How many? _____

45. Have you adopted children? No
Yes: How many? _____

46. If you have had children, how many of them have had DMD? _____

Contraception

47. Do you use contraception? No
Yes: What method(s)?

48. If you have conceived, were the pregnancies planned? No Yes

49. Have you used prenatal diagnosis? No Yes

50. Have you used *in vitro* fertilization and preimplantation diagnosis? No Yes

51. Have you attempted sex-selection? No
Yes:
___ by 'natural' techniques designed to encourage the conception of females
___ by artificial insemination of sorted sperm
___ through sex-determination of fetus and subsequent abortion of all males
___ through prenatal testing and subsequent abortion of affected males
___ by *in vitro* fertilization and implantation of females

52. What were the results of the methods you have used?

53. Have you terminated a pregnancy? No
Yes: How many times? _____

Please use the space below if you would like to add any comments:

APPENDIX II

SUMMARY OF SELECTED DATA

SUMMARY OF SELECTED DATA

	Do you consider yourself to be a carrier of DMD?											
Survey Item	No—do not carry the faulty gene				Yes—do carry the faulty gene				Maybe/Not Sure—about carrier status			
Statistic	Mean	S.D.	Freq.	Percent	Mean	S.D.	Freq.	Percent	Mean	S.D.	Freq.	Percent
Age	40	6.2	34	30.6	42.1	8.7	65	58.6	39.7	8.7	12	10.8
20 – 24	21	-	1	2.9	22.7	0.6	3	4.6	23	-	1	8.3
25 – 34	32	3.5	5	14.7	32.6	1.1	5	7.7	31	2.8	2	16.7
35 – 44	40.3	2.8	19	55.9	39.7	2.2	36	55.4	39.2	3.4	5	41.7
45 – 54	46.2	1.9	9	26.5	48.8	2.7	16	24.6	48.8	2.2	4	33.3
DMD in previous generations	-	-	5	14.7	-	-	18	26.9	-	-	1	8.3
At least one child with Duchenne	-	-	29	85.3	-	-	60	92.4	-	-	11	91.6
1. My friends know that I am, or may be, a carrier of the muscular dystrophy gene.	2.30	2.37	30		4.95	1.64	66		5.17	2.18	12	
2. I keep my carrier status a secret from others.	0.26	0.87	31		0.89	1.64	66		0.45	1.04	11	
3. I feel (felt) I must tell a prospective mate about my carrier potential.	4.12	2.71	25		5.44	1.33	55		4.20	2.90	10	
4. I would like to discuss my feelings about having babies with a counselor of some kind.	1.27	2.10	30		1.32	2.03	62		1.00	1.61	11	
5. I would like to become pregnant and have a baby.	1.72	2.57	32		1.52	2.29	65		1.64	2.11	10	
6. Because of DMD, other people have tried to keep me from having babies.	0.65	1.25	31		0.97	1.80	64		1.08	1.93	12	
7. I have tried to find out whether I am a carrier of DMD through laboratory testing.	5.03	2.05	33		4.77	2.23	66		3.00	3.13	12	

	Do you consider yourself to be a carrier of DMD?											
Survey Item	No—do not carry the faulty gene				Yes—do carry the faulty gene				Maybe/Not Sure—about carrier status			
Statistic	Mean	S.D.	Freq.	Percent	Mean	S.D.	Freq.	Percent	Mean	S.D.	Freq.	Percent
8. Generally, telling people that I am (might be) a carrier has been a mistake.	.41	1.22	27		.70	1.36	66		.67	1.07	12	
9. Being a (potential) carrier makes me unworthy to reproduce.	1.00	1.75	27		1.98	2.27	64		1.42	2.07	12	
10. I talk easily to people about my carrier status.	4.36	2.20	28		4.09	2.07	67		4.67	1.67	12	
11. Being a (potential) carrier puts me at a disadvantage in finding a mate.	.96	1.49	25		1.69	1.93	58		.73	1.68	11	
12. Bearing children is an important part of a meaningful life for a woman.	4.58	1.42	33		4.28	1.85	67		4.33	2.10	12	
13. When I think of being a parent, I think of rearing children, not bearing them.	3.48	2.32	33		3.34	2.14	64		2.50	2.28	12	
14. I have confidence in the accuracy of carrier testing.	4.44	1.81	34		4.63	1.81	67		3.50	1.73	12	
15. I belong or have belonged to a genetic support group.	0.03	0.17	33		0.88	1.85	67		0.50	1.73	12	
16. Experiencing pregnancy is very important to me.	5.16	1.48	32		4.38	2.14	63		4.67	1.88	12	
17. It would be sad to die without having any children to take my family blood line into the future.	3.36	2.26	33		3.58	2.17	64		4.00	2.17	12	
18. I have been told "You shouldn't have children."	0.50	0.95	32		1.48	2.05	66		1.42	2.35	12	
19. I know people with whom I can discuss my feelings about genetic risk.	4.19	2.25	32		4.52	1.78	67		4.17	1.90	12	

	Do you consider yourself to be a carrier of DMD?											
Survey Item	No—do not carry the faulty gene				Yes—do carry the faulty gene				Maybe/Not Sure—about carrier status			
Statistic	Mean	S.D.	Freq.	Percent	Mean	S.D.	Freq.	Percent	Mean	S.D.	Freq.	Percent
20. Being a (potential) carrier makes me less desirable as a mate.	1.15	1.77	27		1.89	1.99	64		1.58	2.11	12	
21. If in a relationship: My mate wants me to have a/another baby.	1.32	2.30	31		0.86	1.72	63		2.09	2.43	11	
22. I am satisfied with my decisions about pregnancy so far.	5.09	1.67	32		5.03	1.38	66		5.33	0.99	12	
23. I feel/felt I should tell a potential mate about my carrier risk…[how soon]	5.20	1.78	25		5.25	1.27	55		5.00	1.50	9	
24. Rate the following roles as to how important you feel they are in your life at present. [total weightings]	37.00	8.71	34		36.32	6.52	66		37.92	9.92	12	
32. Have you seen a Genetic Counselor about whether you are a carrier or not?	0.61	0.50	31		0.55	0.50	62		0.55	0.52	11	
37. Communication to you about pregnancy [total messages]	1.82	3.01	34		3.01	4.89	67		1.50	3.73	12	

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