# Decision Making in Expanded Newborn Screening: The Case of Duchenne Muscular Dystrophy

by

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| | |
|---|---|
| ANOVA | analysis of variance |
| CDC | Centers for Disease Control and Prevention |
| CFIP | Concern for Information Privacy instrument |
| CI | confidence interval |
| DMD | Duchenne muscular dystrophy |
| EHR | electronic health records |
| HBM | Health Belief Model |
| MBSS | Miller Behavioral Style Scale |
| MTurk | Mechanical Turk |
| NBS | newborn screening |
| NCBDDD | National Center on Birth Defects and Developmental Disabilities |
| NHIS | National Health Interview Survey |
| OR | odds ratio |
| SACHDNC | Secretary's Advisory Committee on Heritable Disorders in Newborns & Children |
| SD | standard deviation |
| TRA | Theory of Reasoned Action |

## ABSTRACT

*Background.*

Newborn screening (NBS) is a mandatory public health program aimed at the early identification of babies with conditions that will benefit from early diagnosis and treatment. With increasing technology, some mandatory NBS programs have expanded to offer optional NBS for diseases for which there is limited treatment efficiency data. Little is known about the significant variables that influence parents' decisions about optional NBS. This dissertation used Duchenne muscular dystrophy (DMD) as an exemplar to address whether variation in the presentation and characteristics of NBS tests influence decision making.

*Methods.*

In 3 randomized survey experiments using Internet samples, I explored the following factors that may motivate intended utilization of DMD NBS: 1) the bundling of mandatory NBS panels; 2) the provision of additional information about DMD NBS norms; 3) the mode of DMD NBS results release; 4) the overarching purpose of DMD NBS; and 5) the perceived risk of DMD. The primary outcome variable was intent to utilize DMD NBS with additional outcome variables of attitudes towards DMD NBS. I also explored the influence of these factors on attitudes towards DMD NBS using logistic regressions, and the influence of subjective norms and attitudes towards DMD NBS on DMD NBS intention.

*Results.*

Study 1 findings showed that the presence of a context of mandatory NBS (bundled or unbundled) influenced DMD NBS intent and attitudes towards DMD NBS. When participants were not given the context of broader mandatory NBS in which to place a specific optional NBS, they were more hesitant to choose testing. Presenting additional subjective norm information did not influence DMD NBS intent, though for each study, participants' own subjective norms did predict NBS. Studies 2 and 3 showed that neither the mode of results release nor the overall test purpose guiding the release were significant predictors when parents lacked any specific reason

to believe their child was at risk. However, an interaction of results release and altruism showed that altruistic participants did not choose DMD NBS if their participation would have no societal implications. An interaction of DMD NBS purpose and perceived vulnerability showed that personal purpose increased DMD NBS intent when perceived vulnerability existed, but DMD NBS intent was relatively consistent regardless of perceived vulnerability when the test's main purpose was research. Additional results indicated that medical mistrust is a significant predictor of DMD NBS.

*Conclusions.*

New parents are increasingly being faced with optional NBS decisions, yet there is no consistent policy regarding optional NBS communication, in terms of the information included and the way in which this information is presented. This dissertation explored whether variation in test presentation and characteristics influences optional NBS decision making. The results suggest that future optional NBS programs should be careful to present testing information in a way that explains how a single optional NBS test fits into overall mandatory NBS. Future recruitment in NBS programs should appeal to participants' sense of altruism if, but only if, the participation will, in fact, be a contribution. Additionally, health professionals should attend to parents' perceptions of their child's vulnerability, which appears to be a broader construct than simply family history of a specific disease, as such perceptions influence DMD NBS decision making. Increasing attention to the influence of such structural factors and individual differences on optional NBS decision making will become more and more important as NBS programs are likely to continue expanding.

# CHAPTER I

## Introduction

The time soon after the birth of a child can be a joyful, but often stressful, one for parents. Imagine a couple in their hospital room with their newborn child. The parents are concentrating on their newborn after an exhausting 24 hours with little sleep, and it seems like the nurse has been coming in constantly to talk about medical issues. The nurse now comes in to tell the parents about state-mandated newborn screening (NBS). In addition to this "heel-stick test", the nurse informs the parents about an optional test, one that they can choose to or not to have done on their baby.

The basic differences in mandatory and optional NBS raise questions about how similar, or different, these testing experiences are. While parents are not in a decision-making role regarding mandatory NBS panels, with the continued expansion of NBS, parents may be increasingly faced with the decision to utilize additional optional NBS. However there is no standard practice for delivering NBS information, in terms of the information included and the way in which this information is presented (Hargreaves, Stewart & Oliver, 2005; Loeben, Marteu & Wilfond, 1998). Thus there is a potential for the variation in presentation of and structure of NBS to influence parents' decisions. For example, optional NBS requires separate attention and a conscious decision, even though it is done at the same time as mandatory tests. How optional NBS are presented to parents in the context of mandatory NBS is a potential influence, as is the way in which the results from optional NBS are released. There are multiple ways in which the NBS results can be released, each with varying degrees of accessibility to others. With varied

guidelines for optional NBS, it is important to address these potential determinants; significant influences may lead to non-optimal outcomes such as disconcordance between desire for and utilization of NBS.

**What is Newborn Screening?**

Newborn screening (NBS) is a public health program that allows for early detection of disorders that would cause irreversible clinical damage if not recognized at birth (Therrell, 2001). NBS is a mandatory part of pediatric care, describing a set of laboratory tests including metabolic, hematologic, and endocrinologic tests, as well as genetic analyses. The physical process of NBS begins between 24-48 hours after birth with a single blood sample taken from the baby's heel, which is then dried onto filter paper. The blood is then sent to a laboratory for testing. Results are returned within one week and additional testing is done in the case of positive results.

NBS was first successful in Massachusetts in 1962 with screening for Phenylketonuria (PKU) and serves as a foundation for future NBS (Guthrie & Susi, 1963). PKU is a metabolic disease resulting from an enzyme deficiency. In the 1950s a special diet was developed to prevent the severe retardation that often results from PKU, and a decade later an assay to test for the disease was introduced (Alexander, 2003; Centerwall & Centerwall, 2000). Thus there was a clear justification for PKU NBS: babies benefit from early detection and treatment of the disease. Starting in the mid-1970s, NBS panels expanded to include other genetic disorders (McCabe, Therrell, Larson & McCabe, 2002) and expanded NBS has seen a high compliance rate (Liebl et al., 2002). Generally, expanded NBS is still limited to conditions that benefit from early diagnosis and early treatment (Centers for Disease Control National Center on Birth Defects and Developmental Disabilities [NCBDDD] 2004).

The NBS expansion was significantly aided by the development of tandem mass spectrometry technology, which made it possible to screen for multiple disorders using one blood sample at incremental cost while reducing the rate of false positives (Carroll & Downs, 2006; Insinga, Laessig & Hoffman, 2002; Levy, 1998; Schoen, Baker, Colby & To, 2002). Due to this routinization of tandem mass spectrometry technology, between 1995 and 2005 the average U.S. state added 19 tests for treatable disorders to its NBS program (Tarini, Christakis & Welch, 2006). However there is significant variation in public health programs in the U.S. and internationally regarding the number and types of conditions for which NBS is conducted (Clayton, 1999; Comeau et al., 2004; McCabe et al., 2002; Therrell Johnson & Williams, 2006). In response to this variation, in 2005 the American College of Medical Genetics argued for a uniform NBS panel with 29 core disorders and 25 additional secondary disorders to be used in newborn screening programs in all U.S states (The American College of Medical Genetics, 2006). Despite this report, each state has an NBS program with a panel of mandatory tests that screen for anywhere from 30 to 55 conditions (National Newborn Screening and Genetics Resource Center, 2012). These variations are due to test efficacy, program support, and local advocacy (Moyer et al., 2008).

While NBS programs are widespread, one barrier in offering consistent programs is the significant divisions in how geneticists approach NBS. One side of stakeholders views NBS as a successful example of public health that should be continued as a population-level public health service (Green, Dolan & Murry, 2006; Marsden, Larson & Levy, 2006), while the other side desires evidence-based research to guide NBS education practices (Hoff, Hoyt, Therrell & Ayoob, 2006; Kenner & Moran, 2005; Sewell, Gebhardt, Herwig & Rauterberg, 2004) and more evaluation research in order to offer an evidence-based NBS test, especially for tests that do not

offer treatment implications and should be voluntary (Botkin et al., 2006; Dhondt, 2007; Ross, 2006). This division led to recent panel of experts headed by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children ([SACHDNC], 2012) which recommended that a specific NBS should be considered for addition to mandated NBS panels only if the following conditions are met:

1) The disease can be identified at a period of time (24 to 48 hours after birth) at which it would not ordinarily be clinically detected.
2) A test with appropriate sensitivity and specificity is available.
3) There are demonstrated benefits of early detection, timely intervention, and efficacious treatment.

Despite the SACHDNC criteria, NBS policy is not settled. NBS programs are likely to continue expanding, increasingly including diseases for which limited information about treatment efficacy is available at minimal incremental cost (Carroll & Downs, 2006; Insinga et al., 2002; Schoen et al., 2002).

**What is Optional Newborn Screening?**

The basic objective of NBS is to identify babies with pre-symptomatic conditions that will benefit from early diagnosis and preventive treatment (Levy & Albers, 2000). But with technological advances, more and more types of genetic testing are becoming available, even if the implications of the results of these tests are unclear. Thus there are two different sets of NBS: mandatory and optional. The core difference of mandatory and optional tests is that there is no or limited data on treatment efficacy of optional NBS, and no improvement in clinical outcomes. Optional NBS is typically available as pilot programs to identify potential biomarkers or treatment for newborn diseases and/or improve future NBS evaluation (Pass et al., 2006), but

often have no immediate implications for those being tested, such as personal behavior changes or medical treatment, depending on the disorder. For example, in 1997 the Massachusetts Department of Public Health developed an optional pilot NBS program for 20 additional disorders including rare metabolic diseases (Atkinson et al., 2001). Mandatory NBS is consistently found to be almost universal (Bradley et al., 1993; Clayton, 2005; Liebl et al., 2002; Therrell et al., 2006), and even optional NBS see utilization rates as high as 90% (Bradley et al., 1993; Campbell & Ross, 2003; Dhondt, 2005). However, debates have arisen with the development of optional NBS.

Ethical concerns about NBS for disorders with no proven treatment efficacy include the use of information and the consent process. The consent process of NBS varies by state and by type of NBS (mandatory or optional). Mandatory NBS does not require a formal consent process and is often done without parental awareness (Campbell & Ross, 2003; Davis et al., 2006); in the recent past only three U.S. states required parents' signed consent (Mandl, Feit, Larson & Kohane, 2002) and an opt-out option exists in 27 states either by verbally refusing screening or signing a waiver refusing screening for religious or other reasons (Mandl et al., 2002). Optional NBS involves discussions between parents and healthcare providers and requires explicit consent from parents. The requirement for consent is important because it means that how a test is offered could affect whether or not is done. For example, one criticism offered by new parents is that physicians often inform them about optional NBS decisions just after birth, when they are tired and distracted, instead of before birth, when they would have had a greater opportunity to learn about the disease and the NBS process (Campbell & Ross, 2003).

The combination of advancing technology and parent and provider advocacy has pushed for an increase in optional NBS (Pass et al., 2006). However questions arise about the

presentation and characteristics of specific NBS tests, and they influence of these attributes on choice and attitudes.

**How is Optional NBS Presented in the Context of Mandatory NBS Panels?**

Optional NBS is generally not done in isolation; it is done in conjunction with mandatory NBS panels. For example, in the Massachusetts screening program parents are asked to participate in optional NBS before the blood specimen is sent to the lab for mandatory NBS (New England Newborn Screening Program). The parents' participation is recorded on the blood specimen collection form (parents received a copy of the form). It is crucial, then, to know whether the coupling of optional NBS with mandatory NBS influences parental decision making.

When presented with multiple pieces of information people value each piece less than they do the same information presented in an overall cluster, clustering information increasing the value of that information (Koszegi & Rabin, 2009). Researchers have asserted that people prefer one piece of clustered information versus multiple pieces of information (Koszegi & Rabin, 2009). This clustering is similar to bundling, the strategy of marketing two or more products or services in particular combinations (Wilson, 1997). Bundling is most commonly observed in the marketing of complementary products, such as a cable TV + internet plan versus separately bought plans (Venkatesh & Mahajan, 2009). NBS can be seen as a situation in which multiple products exist – optional and mandatory NBS. In this case there are two ways of presenting NBS to parents: 1) unbundled (presenting each NBS test separately); and 2) bundled (presenting the NBS tests as one single product).

Bundling may affect the utilization of optional NBS by manipulating the attention given to the optional tests versus the mandatory NBS test panel. A qualitative study found that when parents went through a separate invitation process for an optional NBS test (as opposed to a

single occasion that included the optional NBS test offer at the same time as the mandatory NBS tests), utilization of NBS was affected (Moody & Choudry, 2011). This concept of separating the optional and mandatory NBS steps may also apply to the context of mandatory NBS alone; a similar effect may be seen in the bundling/unbundling of mandatory NBS test panel. Additionally, what has previously been described as 'sequential location' (Pilnick, 2008), or the way a NBS test is closely related in proximity to other more routine tests, can be thought of as bundling. Because optional tests are presented with a panel of mandatory NBS tests, an unbundled mandatory NBS test panel may divide the decision makers' attention and make it harder to focus on the single decision at hand – the optional NBS.

**What Happens to the Information from NBS?**

When making the decision to utilize optional NBS, parents may also take into account what happens after the test: what happens with the results and how much control do they have over that information? Currently, NBS results (mandatory or optional) information is typically given to medical professionals, who then share the results with parents and enter them in medical records that parents can access. In 46 states the results from mandatory NBS are released to the birth hospital (Mandl et al., 2002), however information from mandatory NBS results is available to multiple other sources, which has implications for the usage of this information in the future. Parents often report not knowing that the mandatory NBS testing process typically involves state health departments instead of a private laboratory or the hospital laboratory and feeling "blindsided" by that knowledge (Davis et al., 2006). But increasingly, such as in California, private sector laboratories are becoming involved in NBS (California Department of Public Health, 2010). Although these laboratories are public-private partnerships that involve the state-run mandatory NBS programs, there also exist for-profit private genetic testing companies that

can perform optional NBS and provide these results to individuals (McCandless, 2004; Tarini, 2007). NBS information (either mandatory or optional) can also be used for treatment and research. This information is typically collected from residual blood specimens and stored in biobanks. Finally, numerous registries allow such NBS information to be tracked for public health purposes. Each optional NBS release policy may inform parents' anticipated control over the test results, which may in turn influence their decision to utilize the test in the first place.

In the case of optional NBS, pediatricians (or the primary medical care giver) and parents usually have access to the information regardless of whoever else might see it. However, sometimes NBS information is released privately, which grants parents complete control over the information, including whether, and with whom, to share it. Although there is no research addressing the patterns of patients sharing privately-released genetic information with their physicians, previous research has addressed influences on, and consequences of disclosing such information to other family members for whom the results also have implications. Research about the social and psychological consequences associated with revealing the inheritance of a genetic disease has shown that sharing test results can increase tension between family members (Metcalfe 2008) and worry about future children's health (Weil, 2002). Family members are sometimes blamed for genetic disabilities (James et al., 1996), often by the other parent (Weiss, 1981). Bailey et al addressed a deeper ethical issue about disclosing genetic test results to family members - the lack of their explicit informed consent when the test results may present unwanted information about themselves (Alpert, 2003; Bailey, Jr. et al., 2009).

Once the NBS information is released to physicians, the test information is often entered into an electronic health records (EHR) accessible to current and future medical providers, thereby increasing care coordination. EHRs are also meant to give patients more control over

their medical information because they are able to access that information themselves (Wynia & Dunn, 2010). EHRs have general public support: recent studies have shown that approximately 87% of Americans believe that EHRs could improve medical care quality and efficiency (Westin & Markle Foundation, 2008) and almost 50% express interest in using one, even stating that they would pay to use EHRs (Vishwanath, 2009). However in 2010 only 2.7% of people were actually using an EHR (Westin & Markle Foundation, 2008) and privacy concerns are credited with preventing common utilization (Vishwanath, 2009; Westin & Markle Foundation, 2008). These privacy concerns focus mainly on sharing medical test results (Ball, Smith & Bakalar, 2007), and are partly due to the unease over others accessing and using data from EHRs to discriminate against (Grossman, Zayas-Caban & Kemper, 2009).

Privacy concerns may be even more pronounced with NBS: the parent is acting as a proxy decision maker and the test results have long-term implications for other family members who may be worried about future discrimination by health insurers (Tabor et al., 2011). Currently, NBS results are most often given directly to the physician and he or she is responsible for reporting them to the parents. Although patients' EHR utilization is low, the push to move personal health records to an electronic system is increasing. Therefore it is worth considering what might happen if NBS results were automatically entered into an EHR by the physician, an unstudied matter thus far. The concerns that accompany the use of EHRs may then influence the decision whether or not to utilize NBS, making the information from the test visible to medical professionals.

NBS programs often utilize biobanks of residual blood specimens (RBS); 14 NBS programs that serve almost half of all U.S. births save RBS for at least 21 years (Therrell & Hannon, 2012). For example, Michigan maintains a statewide biobank of newborn blood spots

which have been collected and archived on all live births in the state since 1985 (The Michigan Neonatal Biobank). In the case of RBS the control a parent has over the information from that particular NBS test is loosened because he/she typically does not know what that RBS would be used for. The use of RBS for research is a controversial topic. Most studies have found that parents are willing to let RBS be used for research, especially if permission is explicitly obtained (Botkin et al., 2012; Davey, French, Dawkins & O'Leary, 2005; Tarini et al., 2010), and an overall belief in research is cited as a reason for accepting NBS (Parsons, Israel, Hood & Bradley, 2006). Previous research has revealed five influences on parental support for research using RBS: 1. Avoiding harm to their child; 2. Time and convenience; 3. Altruism; 4. Participation incentive; and 5. Relevancy of the study to their family (Tabor et al., 2011). There are also barriers to this support for research, including automated storage processes without explicit permission from parents (Botkin et al., 2012; Tarini et al., 2010) and distrust in authorities (Bombard et al., 2012), which have led to recent lawsuits challenging the storage of RBS (Bombard et al., 2012).

In 2000 The American Academy of Pediatrics Newborn Screening Task Force set forth a broad agenda for state NBS systems. States were urged to develop and support information systems capable of tracking, among many things, long-term outcomes of children with special health care needs who were identified through the NBS program (Newborn Screening Authoring Committee, 2008). Contrasting the full control over information from privately released test results, information from NBS can be made widely available by entering such results into an information system, or registry. A medical registry has been defined as, "…an organized system for the collection, storage, retrieval, analysis, and dissemination of information on individual persons who have either a particular disease, a condition (e.g., a risk factor) that predisposes to

the occurrence of a health-related event, or prior exposure to substances (or circumstances) known or suspected to cause adverse health effects" (United States Department of Health & Human Services). Promising research results and possible clinical trials have brought a need for detailed NBS registries. NBS registries allow for collecting, viewing or searching data regarding patients' phenotype and genotype profiles and other medical information that might benefit future studies and possibly inform new therapies (NCBDDD, 2004).

Registries include both disease-specific and population-specific registries. Disease-specific registries can act as surveillance programs to collect, view, and track both patient genetic data and health events over time to identify future therapeutic strategies and/or research studies (Botkin, Anderson, Staes & Longo, 2009). Disease-specific registries can also provide a forum for patient information exchange and support (Botkin et al., 2009). For example, the Cystic Fibrosis Patient Registry is a 40-year old registry containing more than 26,000 people with Cystic Fibrosis. Through this registry, researchers can study effective treatments, design clinical trials, and follow trends in patients' health status (The Cystic Fibrosis Foundation, 2011). Broader, population-specific registries monitor segments of the population, tracking trends in health and healthcare. These registries, typically state- or county-wide, are used to report on gaps in healthcare and inform healthcare services policy. For example, KIDSNET is a Rhode Island Department of Health registry that follows the health and well-being of children up to age 18 (Rhode Island Department of Health). Despite the existence of numerous NBS registries, there has been no research into whether parents' NBS decision making is influenced by the amount of control over the use of NBS information in registries.

**Summary**

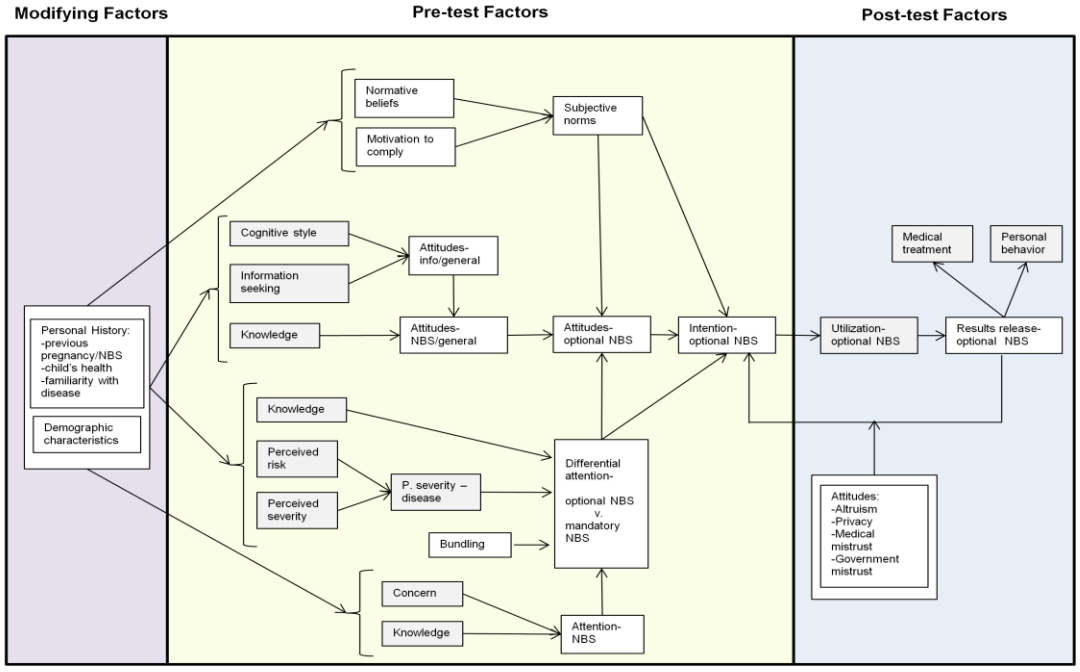
NBS is itself not a new concept. Since the 1960's public health programs have been screening infants for serious diseases that have benefits to early diagnosis, namely an effective treatment. But with the expansion of optional NBS this area has become more complex; optional NBS is intermittently offered and, unlike mandatory NBS, provides no information with clinical implications. The decision to utilize optional NBS is left to the parents. While technological advances and optional NBS become more available and less expensive, there is a potential for increased optional NBS use. It is important to consider how these tests are being presented to decision makers before policies and universal guidelines are set, as the presentation and structure of optional NBS may influence utilization. Knowing whether such decisions are affected by these features may therefore inform future optional NBS practices.

## CHAPTER II

### Theoretical Framework

The focus of this dissertation is to test how optional test decision making varies by features of test presentation and structure, as represented in optional newborn screening (NBS). I placed optional NBS decision making within the theoretical contexts of the Health Belief Model (HBM) and the Theory of Reasoned Action (TRA), with additional constructs informed by previous NBS research. The figure below is the theoretical framework I developed to connect existing work and guide future research (See Appendix A for a larger image). The framework is divided into three sections: modifying factors, pre-test factors (or influences that occur before NBS), and post-test factors (or influences that occur after NBS). Constructs not analyzed in the dissertation studies are shaded in grey.

**Figure 1. Theoretical Framework**



**Modifying Factors**

The modifying factors included in the theoretical framework above are informed by HBM. HBM describes modifying factors as the sociodemographics, personal experience/history and/or underlying knowledge that influence one's health perceptions (Champion & Skinner, 2008). The role of sociodemographics has been studied in nearly every testing context, except for NBS. Personal history has been a particularly influential modifying factor in screening decisions. In prenatal screening, personal experience with pregnancy, health-related matters, genetic conditions and disability affected interest in genetic screening (Archibald & McClaren, 2012; Etchegary et al., 2008). Although NBS has somewhat different implications from prenatal testing, they share similar influences of personal history and experience. Previous NBS research has shown that personal history, such as previous pregnancies and experience with genetic conditions, influences NBS decision making and whether parents feel that those decisions are informed (Davey et al., 2005; Lipstein et al., 2010; Nicholls & Southern, 2012).

**Pre-test Factors**

The pre-test factors in the theoretical framework are based largely on TRA (Fishbein & Ajzen, 1975), which has three main components: behavioral intention, attitudes, and subjective norms. A main tenet of TRA is that the main predictor of behavior is behavioral intention, which measures a person's strength of intention to perform a specific behavior. Therefore in this framework, the predictor of *optional NBS* is *optional NBS intention*. TRA also asserts that behavioral intention is a function of the other two theoretical constructs, attitudes and subjective norms towards the specific behavior. The theoretical framework above presents these TRA pathways.

Turning to attitude constructs, the *attitudes towards the optional NBS* are influenced by *attitudes towards NBS in general*; though exact views may change from test to test, how one views NBS overall (important, unethical, etc.) informs the views about a specific NBS. The *attitudes towards NBS in general* are, in turn, likely informed by two constructs: *knowledge about NBS* and *attitudes towards information in general*. For example, there is the belief that the more information a person has, the better; this attitude would indicate support of NBS which gathers information about a child. *Attitudes towards information* have strong cognitive influences, such as *cognitive style* and *information-seeking* style. The need for cognition, or the tendency to engage in cognitive activity, is a type of cognitive style that has been shown to influence health beliefs and medical decision making (Cacioppo, Petty & Kao, 1984). Monitoring or blunting, or the tendency to seek or avoid threatening information (Miller, 1987), is one way to describe the way of seeking information. The Monitoring and Blunting Coping theory posits that people differ in their preference for information during an unfavorable event. This "difference" guides peoples' attitudes towards information. The "monitors" are proactive and seek; they want to know all of the current information because it will alleviate stress and uncertainty. The "blunters" are passive and avoid information; they do not seek to alleviate uncertainty and prefer a state of ignorance if there is a risk of undesirable information/outcomes. Both of these types are seen in parents making NBS decisions, with some monitors actively seeking out information, and others only learning about NBS incidentally (Tluczek, Orland, Nick & Brown, 2009). In fact, one study found that parents classified as monitors requested information about the nature and purpose of NBS, as opposed to technical details or aspects relating to prevalence (Campbell & Ross, 2004).

The final element of TRA is subjective norms. This is an important factor in the theoretical framework, directly influencing NBS intention. Subjective norms are a consistent theme in NBS decision making; parents use a variety of sources of information and decision support in their NBS decisions. As developed in TRA, subjective norms are determined by one's *normative beliefs* (whether a relevant person approves or disapproves of a behavior), weighted by his or her *motivation to comply* with that person (Montano & Kasprzyl, 2008). NBS programs provide information and decision support through pamphlets, pediatricians, midwives or nurses (Hargreaves et al., 2005; Munck et al., 2007; Parsons et al., 2006), but in addition to these medical sources parents rely on the experiences of their friends and family members (Davey et al,. 2005). Experiential knowledge is an important source of information for parents, which includes one's own personal experience as well as the knowledge gained from others (Etchegary et al., 2008). In fact, much of parents' NBS knowledge comes from hearing other parents at newborn visits (Detmar et al., 2007). It is not just NBS information receipt that is affected by family and friends; in a Welsh study of optional NBS utilization, the influence of family's negative experience was the cause of higher refusal rate in a particular study area (Bradley, Parsons & Clarke, 1993). Subjective norms also indirectly influence intention with attitudes towards optional NBS as a mediator.

The *differential attention paid to the specific optional NBS v. mandatory NBS* presented plays an important role. This describes the difference in attention a person pays to a single NBS test in the context of a mandatory NBS panel present, based on how that mandatory NBS panel is *bundled*. With a bundled mandatory NBS panel, there is a greater opportunity to focus on the single optional NBS decision. Mandatory NBS panels that are *unbundled* will directly affect the differential attention by presenting the mandatory tests individually, versus presenting

information as a group or a single unit. This *differential attention* directly influences *optional NBS intention*. Differential attention also affects the *attitudes towards specific optional NBS*, by pulling focus from, for example, the importance of the test.

There are additional influences on both attention and differential attention, including *perceived severity of the disease (*a combination of *perceived severity* and *perceived risk)*, and *concern* and *knowledge* about NBS. Perceived severity of the NBS for which the NBS is being done is constructed of both perceived risk and perceived severity, and is deeply influenced by one's personal experience and familiarity with the disease (Davey et al., 2005). Depending on one's perceived severity, he/she might pay less or more attention to the decision at hand. With experience and exposure to NBS one builds knowledge about the topic. Although studies have suggested that overall parents have a poor knowledge of NBS topics (Tluczek et al., 1992; Tluczek et al., 2005), increased knowledge is associated with increased attention to NBS.

**Post-test Factors**

In addition to the factors that occur before the NBS takes place, there are post-test factors that affect the decision to utilize optional NBS. In other words, what will happen after NBS is done influences whether someone has NBS in the first place. In the NBS timeline, after NBS is completed the next action step is the *release of NBS results.* The information from these results can be used for *medical treatment* or change in *personal behavior,* depending on the disorder and the treatment available, which are not universally proven to be effective but can be offered through optional NBS pilot programs.

As shown in the theoretical framework, there is a feedback loop from releasing NBS results to NBS intention – knowing what will happen to the information may influence whether a person initially chooses NBS. Previous work has shown that parents have opinions about the use

of NBS information (Tabor, 2011); these opinions may also inform NBS intent. This feedback loop is moderated by a number of attitudes, however. First, one's *attitudes towards information*; the theoretical constructs that contribute to these attitudes are cognitive style, such as need for cognition (Cacioppo et al., 1984), and information seeking, such as monitoring or blunting coping (Miller, 1987). For example, those who value having and sharing information will be more likely to utilize optional NBS if the results are to be disseminated widely.

Attitudes towards *altruism*, *privacy*, *medical mistrust*, or *mistrust in government* are also influential moderators. There is a long history in the United States of medical mistrust, especially among racial minorities (Corbie-Smith, Thomas, Williams & Moody-Ayers, 2002; Corbie-Smith, Thomas & St. George, 1999). One study found that trust in the medical community was central to the attitudes of the mothers considering NBS (Parsons et al., 2006). As outlined above, these three concepts have been shown to influence parents' opinions towards NBS. For example, those who are altruistic and believe in helping society overall, do not have overwhelming privacy concerns about personal information, or do not mistrust the medical system, will be less likely to have their optional NBS decisions swayed by the way in which information is to be used.

**Theoretical Framework Summary**

The determinants of optional NBS decision making can be described in three different stages: 1) modifying factors such as sociodemographics, relevant personal experience/history (e.g. pregnancy), and underlying knowledge that influence one's health perceptions; 2) pre-test factors such as NBS intention, informed largely by TRA as a function of attitudes and subjective norms, and differential attention; and 3) post-test factors that will occur after NBS is done, such as the release of NBS results. The feedback loop in the theoretical framework emphasizes how important knowing what will happen in future is to the initial decision. The individual

characteristics and personal attitudes included in the framework, such as attitudes towards altruism, privacy, and mistrust in the healthcare system, complete a NBS decision making process influenced by the presentation of testing information and characteristics of the specific test.

# CHAPTER III

## An Exemplar: Duchenne Muscular Dystrophy

The focus of this dissertation is to show that optional testing varies by features of test presentation and structure. Previous newborn screening (NBS) decision making research has focused mainly on the technical process of NBS and the psychosocial outcomes of NBS, but with the rise of optional NBS there is a need to study the specific determinants of these decisions. Detailed exploration of NBS decision making requires consideration of the details involved in a specific NBS context. This dissertation uses the exemplar of NBS for Duchenne muscular dystrophy (DMD). The larger research questions relevant in optional testing are specifically important in DMD NBS decision making.

### An Introduction to Duchenne Muscular Dystrophy

DMD is a rare form of muscular dystrophy. It is a lethal X-linked genetic disorder for which a defective gene for a muscular protein causes rapid muscular degeneration. DMD can occur in people without a known family history (Kleigman, Behrman, Jenson & Stanton, 2007). Typically DMD occurs in boys; there is an incidence of 1 in 3,500 boys worldwide (Duchenne Muscular Dystrophy Research Fund). Although there are preliminary trials testing the use of steroids for DMD symptoms (ClinicalTrials.gov, 2011) no treatment is widely used that can alter the disease course of DMD. Therefore learning one's DMD status does not improve clinical outcomes. Typical onset is between 3-5 years of age; symptoms include fatigue, learning difficulties, and muscle weakness (Ciafaloni et al., 2009). The muscle weakness is so progressive

that the majority of boys are in wheelchairs by the age of 10, have breathing difficulties and heart disease by age 20, and survival beyond 30 is rare (Kleigman et al., 2007; NCBDDD, 2004).

**Duchenne Muscular Dystrophy Newborn Screening**

In the 1970s, researchers developed a NBS blood test to identify cases of DMD (NCBDDD, 2004). The test was not introduced into the mandatory NBS panel because it provided no effective clinical advantages (Ross, 2006). Although DMD NBS is currently not available systematically, single pilot programs have offered the screening. Table 1 presents a brief history of pilot DMD NBS programs (Mendell et al., 2012), beginning in New Zealand (Drummond, 1979) and most recently in Ohio (Mendell et al., 2012). This most recent program in Ohio, the Statewide Newborn Screening for Duchenne Muscular Dystrophy program , offered voluntary DMD NBS from 2007-2011 in conjunction with The Research Institute at Nationwide Children's Hospital, Cincinnati Children's Hospital and the Ohio Department of Health, The University of Utah, and the Centers for Disease Control. DMD NBS was offered initially through a pilot study in several birthing hospitals in Columbus and Cincinnati, followed by an expansion to birthing hospitals throughout the state. This DMD NBS program was developed as a template to be expanded to a national voluntary screening program. DMD NBS utilization was high for all of the pilot DMD NBS programs (Parsons et al., 2006), with only one program still continually offering DMD NBS, in Antwerp.

**Table 1. A History of DMD NBS**

| Year of Report | Country | Investigators | Number newborns screened |
|---|---|---|---|
| 1979 | New Zealand | Drummond L.M. | 10,000 |
| 1982 | Edinburgh, UK | Skinner R., Emery A.E.H., Scheuerbrandt G., et al. | 2,336 |
| 1986 | West Germany | Scheuerbrandt G., L€ovgren T., Mortier W. | 358,000 |
| 1988 | Manitoba, Canada | Greenberg C.R., Jacobs H.K., Nylen E., et al. | 54,000 |
| 1989 | Lyon, France | Plauchu H., Dorche C., Cordier M.P., et al. | 37,312 |
| 1991 | Western Pennsylvania, USA | Naylor E.W. | 49,000 |
| 1993 | Wales, UK | Bradley D.M., Parsons E.P., Clarke A.J. | 34,219[a] |
| 1998 | Cyprus | Drousiotou A., Ioannou P., Georgiou T., et al. | 30,014 |
| 2006 | Antwerp, Belgium | Eyskens F., Philips E. | 281,214[b] |
| 2012 | Ohio, USA | Mendell J.R., Shilling C., Leslie N.D., et al. | 30,547 |

[a]A second presentation in 2011 reported 335,045 newborns screened
[b]Only DMD NBS program continually active

The arguments against DMD NBS are broad. In general, NBS must address the psychosocial, clinical and reproductive implications of genetic information for the child and the family (Ross, 2006); it is argued that optional NBS like DMD NBS, which expands information beyond the immediate care of the child, "muddles" the primary purpose of NBS (Bailey et al., 2008). There is an overall desire "not to know" DMD NBS results (Parsons et al., 2006). Parents have reported that an DMD early diagnosis might lead to earlier stigmatization and discrimination (Dhondt, 2010), and diagnosing an illness without clinical treatments may disrupt the parent/newborn bonding relationship (Bailey Jr. et al., 2009; Goddard & Cardinal, 2004). Parents refusing DMD NBS have also cited the lack of treatment, no direct health benefits or economic value as their motive (Campbell & Ross, 2003; Parsons et al., 2006; Whitehead, Brown & Layton, 2010). In one study, 22% of parents refusing DMD NBS described no real

benefit of knowing and 14% cited no cure or proven treatment as their reason for declining (Cyrus, Street, Kable, Fernhoff & Quary, 2012). Finally, some parents are unsure of the implications of a positive NBS result (Ciske, Haavisto, Laxova, Rock & Farrell, 2001; Lang, McColley, Lester & Ross, 2011), although initial hyperawareness has not been associated with overuse of health care services (Lipstein, Perrin, Waisbren & Prosser, 2009). These existing drawbacks to DMD NBS have prevented it from becoming part of mandatory NBS.

A CDC-sponsored workshop was held in 2004 to discuss not only the risks of, but also the benefits of DMD NBS. The workshop concluded that although there was inadequate evidence showing medical benefit from DMD NBS, early diagnosis might have other nonmedical advantages (NCBDDD, 2004). An earlier diagnosis could offer knowledge benefits by informing parents' or other family members' reproductive planning (Goddard & Cardinal, 2004; Parsons et al., 2006) or an opportunity for immediate DMD education (Bailey Jr. et al., 2009). Overall, more "time to prepare" has been cited widely as a benefit to DMD NBS, including relocating near treatment centers, home purchasing, financial planning, or employment opportunities (NCBDDD, 2004; Parsons et al., 2006; Pelias, 2006). As there are no universal early signs or symptoms that pediatricians use to recognize young children with DMD, so numerous testing and medical appointments may be necessary until final diagnosis, such as electromyography (EMG), muscle biopsy, or blood tests for CPK, an enzyme (Kleigman et al., 2007). This investigative testing experience is known as the "diagnostic odyssey", and can be a costly and anxiety-provoking one (Cyrus et al., 2012; Lipstein, Brinkman & Britto, 2012). Thus a number of parents see great benefit to an early diagnosis of DMD.

*Exploring Emerging Questions*

The small body of NBS decision making research has been limited to psychosocial outcomes and parents' attitudes towards mandatory NBS. Although it is known that partitioning NBS invitations decreases utilization (Moody & Choudhry, 2011), we do not know if related factors are potential influences on optional NBS decisions. Future optional NBS practices, which will only increase as technology advances, may be informed by knowing whether these decisions vary by features of the test presentation and structure. DMD offers an exemplar to study optional NBS questions because DMD testing occurs outside of the standardized processes of mandatory NBS and requires a conscious decision.

# CHAPTER IV

## Preliminary Study

### Introduction

Little work has studied whether optional testing decisions vary by features of the test presentation or structural characteristics of the test. To start exploring these issues, I conducted a pilot study using DMD NBS as an exemplar of an optional test presented in the context of mandatory NBS. The study looked at this presentation characteristic as well as the test characteristic of test burden using experimentally-manipulated groups. Outcome variables included worry about the DMD NBS results and reported importance about: DMD NBS, DMD NBS results, and information from the DMD NBS results.

### Methods

*Study Participants*

Preliminary data collection used 2,085 adult participants in a larger, Internet-administered decision making study. Participants were recruited using Amazon.com's Mechanical Turk (MTurk) interface. The average age of participants was 49 (SD = 16.3). Overall, participants were white (73.8%) and well-distributed in terms of education (22.5% attended some high school or attained a high school degree, 34.2% attended some college, and 43.3% attained a college degree or higher). Gender was evenly split between female (52.0%) and male (48.0%).

*Research Design*

Upon entering the study participants were assigned to one of three scenarios (see Appendix B for Preliminary Study Vignettes), each of which contained the following

information: you and your partner have just given birth to a baby boy; DMD cannot be cured and the symptoms cannot be prevented; there is an optional test for screening newborn babies for DMD; the screening requires a blood sample but causes no long-term harm; the screening will not cost any money. The study explored the statistical differences in the three experimental groups that varied on three factors: bundling of the mandatory NBS panel presented with the optional DMD NBS, burden of DMD NBS, and the automatic release of DMD NBS information.

Group 1: Participants in this group saw a mandatory NBS panel presented with optional DMD NBS that was bundled (the multiple NBS tests were presented as one package, "49"). The burden of the DMD NBS process, or the amount of logistical burden placed on the parent to complete the DMD NBS and the length of the NBS process, was non-existent; DMD NBS was done in the hospital with an existing blood sample so no additional actions were required. Finally, the information resulting from DMD NBS was to be released automatically.

Group 2: Participants in this group saw a mandatory NBS panel that was unbundled (each of the 49 mandatory NBS in the panel were shown separately). The burden of DMD NBS was moderate (i.e. parents would have to send in a permission form for DMD NBS to be done on the existing blood sample), and the resulting information was not automatically released.

Group 3: Participants in this group read about DMD NBS without the context of mandatory NBS, therefore the bundling factor was not manipulated. The DMD NBS burden was high—parents would have to return to the hospital at a later time for DMD NBS. There was no specific mention of the DMD NBS results; therefore automatic release factor was not manipulated.

*Hypotheses*

H1) Participants given bundled DMD information (Group 1) will be more worried about DMD NBS results than participants given unbundled DMD information (Group 2).

This hypothesis tested the influence of bundling the mandatory NBS panel presentation. Based on the bundling literature, viewing the bundled (vs. the unbundled) mandatory NBS panel would allow participants to attend more to the single DMD NBS decision. This increased attention would lead to increased worry; participants who focused less on that single NBS decision would subsequently worry less about its specific results.

H2) Participants given a high DMD NBS burden (Group 3) will report the DMD NBS, DMD NBS results, and the subsequent information as less important than participants given a low DMD NBS burden (Group 1).

This hypothesis tested the influence of the time and effort required for DMD NBS on the value placed on DMD NBS and its results. With an increased burden people would see all facets of DMD NBS as less important and not worth the time and extra effort.

**Results**

The first hypothesis proposed that participants in Group 1, those shown a bundled mandatory NBS panel, would be more worried about DMD NBS results than participants in Group 2, those given an unbundled panel. As hypothesized, Groups 1 and 2 differed significantly on their reports of worry about the DMD NBS results (see Table 2). Though there were confounding factors, this result suggests that Group 2 gave less attention to DMD NBS compared to Group 1, and therefore focused less and worried less about that specific NBS result.

**Table 2. Group Differences in Reporting Worry about DMD NBS Results**

| | Group 1 - % (N) *Bundled* | Group 2 - % (N) *Unbundled* | $Chi^2$ |
|---|---|---|---|
| Worry about DMD NBS results | 52.72 (242) | 43.81 (216) | 12.48 ($p<0.05$) |

The second hypothesis proposed the participants in Group 3 (high burden DMD NBS) would report the DMD NBS, DMD NBS results, and the subsequent information as less

important than participants in Group 1 (low burden DMD NBS). As proposed, there was a significant difference between Groups 1 and 3 in the 'importance' outcome variables (see Table 3). It is difficult to tease apart these 2 groups by a specific factor, but knowing that for each of these variables Group 1 more often answered 'more important' may indicate that as the burden and length of the DMD NBS process increased, DMD NBS became less important and the information gleaned from the test was seen as not worth the time and extra effort.

Most of the participants in Group 3, chose whether they would return to the hospital at an additional time for DMD NBS, reported that they would 'definitely' (32.9%) or 'probably' (41.7%) do so. Participants in Group 2, who chose whether they would mail in a permission form to have DMD NBS on an existing blood sample, responded similarly with 35.8% and 39.4% reporting they would 'definitely' or 'probably' do so, respectively (t=0.68, n.s.). Substantial proportions of participants strongly agreed that the DMD NBS results would help them prepare for the future (45.3%), affect whether they had more children (24.2%), and affect how they may treat their child (20.9%).

**Table 3. Group Differences in Reporting DMD NBS 'Very Important'**

| | Group 1 -%(N) *No burden* | Group 3 -%(N) *High burden* | $Chi^2$ |
|---|---|---|---|
| DMD NBS 'very important' | 48.71 (226) | 31.94 (160) | 32.79 ($p<0.001$) |
| DMD NBS results 'very important' | 58.48 (269) | 48.40 (242) | 9.86 ($p<0.05$) |
| Information from DMD NBS results 'very important' | 69.63 (321) | 56.49 (283) | 22.69 ($p<0.001$) |

**Implications for the Design of the Primary Research Studies**

The preliminary study showed group differences in worry, indicating that unbundling the mandatory NBS panels required participants to divide their attention and focus less on the DMD NBS decision. Group differences in the importance of DMD NBS, DMD NBS results, and the

resulting information indicated that as the burden and length of the DMD NBS process increases, the value of the test decreases. However the presence of multiple possible confounding factors suggests the need for a study with a multifactorial design. Therefore, these results support the notion that DMD NBS decisions do vary by features of presentation and structure, and advocate for further study of these concepts.

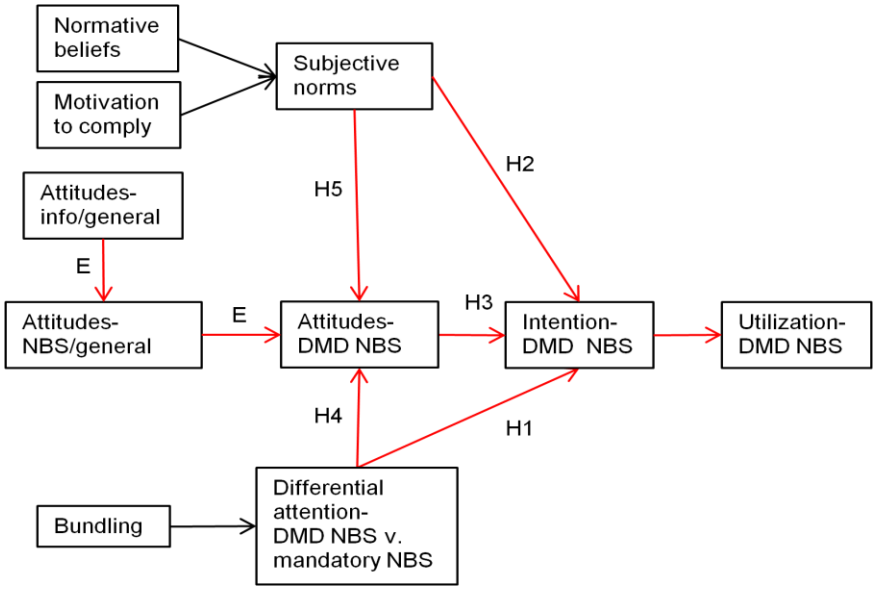
# CHAPTER V

## Study 1

### Introduction

Optional NBS, such as DMD NBS, is often presented with mandatory NBS panels. The overarching research question of this dissertation is whether DMD NBS decisions vary by features of test presentation or structural characteristics of the test. Three studies separate studies addressed this question. Study 1 considered the influences on DMD NBS utilization given the context of mandatory NBS panels, specifically the notion of bundling. Researchers have supported the effect of bundling similar information, but none have studied how one decision is made in the context of bundled information. Study 1 also investigated the effect of subjective norms on the decision to utilize optional DMD NBS. NBS experiences of one's families and friends are very influential in NBS decision making; optional NBS refusal rates have been attributed to family members' negative experiences. In addition to their own experience and medical information, parents' decisions may depend on subjective norms information. The conceptual model for Study 1 (Figure 2, below) describes specific hypothesized influences on the outcome variable *DMD NBS intent.*

**Figure 2. Study 1 Conceptual Model**




I hypothesized that *bundling* the panel of mandatory NBS would predict DMD NBS intention through its manipulation of the *differential attention* paid to the mandatory NBS panels versus the single DMD NBS (H1). In the case of a panel with 49 mandatory NBS, I hypothesized that presenting DMD NBS next to 49 mandatory NBS listed one-by-one (unbundled) would highlight every NBS test and divide one's attention, so participants would be more aware of (and possibly overwhelmed by) every NBS in the panel and be able to focus less on the single decision at hand than those seeing a bundled NBS panel (the statement "49 tests"). Participants viewing unbundled panels would be less likely to intend to utilize DMD NBS, compared to participants viewing bundled mandatory NBS panels. Essentially, the amount of attention that a person can give to DMD NBS would be affected by the way the mandatory NBS tests are shown in the panel, which may draw away attention.

I also hypothesized that *subjective norms* would predict DMD NBS decision-making (H2). From the Theory of Reasoned Action ([TRA] Fishbein & Ajzen, 1975), subjective norms describe beliefs about what others want (*normative beliefs*) and how much people want to

comply with those desires (*motivation to comply*). Showing participants additional subjective norm information about other parents' DMD NBS decisions ("most parents agree to DMD NBS") would increase their intent to utilize DMD NBS. Finally, I hypothesized that *attitudes about DMD NBS* would predict DMD NBS intent, with more positive attitudes about DMD NBS associated with higher intent (H3).

I developed hypotheses to test the influences of the study factors on the outcome variable *attitudes about DMD NBS*. *Bundling* would influence attitudes about DMD NBS by providing different contexts for focusing on DMD NBS (H4). Participants seeing unbundled mandatory NBS panels would report less positive attitudes about DMD NBS. *Subjective norms* would influence attitudes about DMD NBS (H5), as participants would have more positive attitudes as a reflection of the additional information about other parents' high DMD NBS utilization.

I set forth exploratory aims (E) to examine the associations between different attitudes in DMD NBS intention. The conceptual model presents links between *attitudes about information in general*, *attitudes about NBS in general*, and *attitudes about DMD NBS*. I tested this pathway of general to specific attitudes.

*Study Aims and Hypotheses*

Primary Aim: To examine the influences of the experimental factors on intended utilization of optional DMD NBS.

H1: Participants seeing bundled mandatory NBS panels will report higher DMD NBS intention, compared to participants seeing unbundled mandatory NBS panels.

H2: Participants viewing additional subjective norm information about parents' DMD NBS use will report higher DMD NBS intention, compared to participants not seeing such additional information.

H3: Reporting more positive attitudes about DMD NBS will be associated with higher DMD NBS intention

Secondary Aim: To examine the influences on attitudes about optional DMD NBS

H4: Participants seeing bundled mandatory NBS panels will report more positive attitudes about DMD NBS, compared to participants seeing unbundled mandatory NBS panels.

H5: Participants viewing additional subjective norm information about parents' DMD NBS use will report more positive attitudes about DMD NBS, compared to participants not seeing such additional information.

Exploratory Aim: To investigate the associations between different attitudes in DMD NBS intention

**Study Design**

The study had a 3x2 between-subjects experimental design: Bundling (bundled, unbundled, no panel) x subjective norm (specified, unspecified). Participants were randomly assigned to one of the six experimental conditions:

| | | **Subjective Norm** | |
|---|---|---|---|
| | | Norm specified | Norm unspecified |
| **Bundling** | Bundled panel | 1 | 4 |
| | Unbundled panel | 2 | 5 |
| | No panel | 3 | 6 |

Stemming from the literature and the conceptual model, bundling represented how the mandatory NBS tests are presented in a panel. By experimentally manipulating the information presentation in the study materials, participants were randomized into one of the three bundling groups: bundled, unbundled, or no panel. Participants in the bundled group saw a study brochure in which the mandatory NBS test were shown as a package ("49"). Participants in the unbundled

group saw a more detailed brochure in which each mandatory NBS test was shown separately. For those in the no panel group, mandatory NBS was not mentioned in the brochure. Participants were also randomized to a subjective norm group based on manipulation of study materials: norm specified or norm unspecified. Participants in the specified group read information about whether other parents are choosing the optional DMD NBS in addition to the basic information about NBS and DMD. They read the following sentence: "While you are talking, the nurse also tells you that most of the parents that she talked to about this test have agreed to have their baby screened for DMD." Participants in the unspecified norm group did not read any additional information about other parents' decisions.

## Methods

### *Study Population and Participant Recruitment*

The study population was adult, Internet users living in the United States. An Internet survey facilitated the experimentally-manipulated conditions and ensured high internal validity of the randomized experiment by limiting selection bias. In a study with experimentally-manipulated factors with a focus on variations across versions, my goal was to support internal validity. Participants were recruited using Amazon.com's Mechanical Turk (MTurk) interface, which collects anonymous data for experimental studies. There were many advantages to using MTurk. First, MTurk survey respondents are often more representative of the U.S. population than in-person convenience samples (Berinsky, Huber & Lenz, 2011). Second, MTurk provides potential participants with the comfort of completing the survey at any time and any place, which increases response rates. Finally, MTurk recruitment is a rapid, cost-efficient method for recruiting participants. I followed established MTurk "tips" for recruitment, such as including the link to the survey in the HIT (human interface task), or survey request (Berinsky et al., 2011).

Participants were paid $0.75 for their participation directly through the MTurk system. This amount is consistent with current psychology lab practices on MTurk, which typically pay $0.10-$1.00 for a 25-minute survey.

*Inclusion/Exclusion Criteria*

Inclusion criteria included: above the age of 21, United States resident, and the ability to complete a web-based survey. Age and United States residency were verified through MTurk user registration. The ability to complete a web-based survey was confirmed through the MTurk system, based on previous user performance. Similar to typical MTurk experimental studies, I enrolled only participants with a 90% "approval rate", or a history of completing at least 90% of started surveys. Exclusion criteria included: under the age of 21, and less than a 90% approval rate.

*Data Collection*

The research program's experimental study design lent itself to quantitative data collection. Qualitative data collection does not present the ability to cleanly manipulate multiple conditions involving visual distinctions, or assess immediate responses that reflect instinctive reactions. To collect such responses I programmed the surveys using the online survey platform Qualtrics.

To begin each study, participants read a vignette that outlined a hypothetical situation in which they have a newborn (see Appendix C for Study 1 Vignettes). The vignette specified a male newborn, because most DMD NBS pilot programs are offered to male newborns only. This is because while DMD NBS identifies all male newborns with DMD, only some female carriers of DMD are detected and of those, full symptom expression of DMD is not common. Thus DMD NBS results are difficult to interpret in female newborns. Participants then read a brochure that

provided a detailed description of DMD, DMD NBS, DMD NBS results (see Appendix D for Study 1 Brochures). The format of information included in this brochure was modeled after the study brochure from the Statewide Newborn Screening for Duchenne Muscular Dystrophy pilot program in Ohio (Mendell et al., 2012; contact author for a side-by-side comparison of the study brochure and the Ohio pilot brochure). Both vignettes and brochures included manipulated study factors based on the experimental design, described above. After reading the vignettes and the brochures participants completed survey items. Items given to participants in Study 1 are described below; additional items for Studies 2 and 3 are described in those study chapters (see Appendix E for full items).

*Measures*

DMD NBS Utilization

In the real world, the primary outcome of interest is utilization of DMD NBS. In the context of this hypothetical research study and informed by the TRA (Fishbein & Ajzen, 1975), I assessed *intent* to utilize DMD NBS as stated by participants. I used the following question: How likely do you think that it is that you will choose to have your baby tested for DMD?. I used a bidirectional, 4-point Likert scale with endpoints labeled Very Unlikely and Very Likely.

Attitudes & Beliefs

Following the conceptual model, I assessed respondents' attitudes and beliefs surrounding three topics: 1) DMD NBS, 2) NBS in general, and 3) information in general (see Appendix E for full items). To assess attitudes about DMD NBS, I asked participants questions surrounding two topics: the importance of DMD NBS and the impact of DMD NBS. First, participants reported how important is…1) DMD NBS; 2) seeing the results of DMD NBS; and 3) sharing the results of DMD NBS. Responses were on a 5-point Likert scale (Not at all

Important-Very Important). Then participants responded to the following five questions/statements about the impact of DMD NBS items, each on a 5-point Likert scale: 1) How much do you think you would worry about the results of your baby's DMD test? (Not at all-Very Much); 2) The information from the DMD test may help me prepare for the future (Strongly Disagree-Strongly Agree); 3) The information from the DMD test would affect whether I have more children (Strongly Disagree-Strongly Agree); 4). The results from the DMD test may affect how I treat my child (Strongly Disagree-Strongly Agree); 5) My child would be treated differently by others if he is diagnosed with DMD (Strongly Disagree-Strongly Agree).

To assess participants' attitudes about NBS in general, they responded to the following two statements, both on a 5-point Likert scale from Strongly Disagree-Strongly Agree: 1) My child be treated differently by others if he has an incurable disease; 2) Having a child with an incurable disease would change how I might treat my child.

Finally, I assessed participants' attitudes about information in general with the following question on a 5-point Likert scale from Not at all Important-Very Important: How important is it that you have all the information available about your child?.

Subjective Norms

I measured participants' own subjective norms about two topics: NBS in general and DMD NBS specifically. I used direct measures of subjective norms around NBS and DMD NBS specifically with the following two questions: 1) Do you think that most people agree or disagree that it is important for babies to be tested for as many genetic diseases as possible?; and 2) Do you think that most people agree or disagree with getting the DMD test?. I assessed the subjective norms on a scale scored from -3 to +3 (Agree – Disagree). There are established indirect measures of subjective norms based on the TRA's normative beliefs and motivation to

comply, however I chose to use these direct measures. The indirect measures were not practical given the study methodology, because participants did not provide information about specific referent individuals in their lives with whose norms they want to comply. For example, an indirect measure of subjective norm would require a participant to name an important person (e.g. a mother) whose opinions/advice the participant wants to follow.

Participant Demographics

I collected the following demographics: gender (male; female), educational attainment (some high school; high school graduate/GED; some college or technical school; college degree; advanced degree), marital status (married/partnered; not married/partnered), and household income. The response options for household income were based on the National Health Interview Survey ([NHIS]; Centers for Disease Control and Prevention [CDC]) response options calculated by adjusted poverty level based on family size: less than $14,500; $14,500-less than $35,000; $35,000-less than $50,000; $50,000-less than $75,000; $75,000-less than $100,000; $100,000 and over. Participants reported their race: White; African American/Black; Native Hawaiian and Other Pacific Islander; Asian; American Indian/Alaska Native. The response options for race were also based on the NHIS (CDC). Participants entered their age in an open-ended question, which was later categorized into: 21-35, 36 and above, missing. I chose 35 as the cut-off point because pregnancy at age 35 is the typical point at which additional prenatal screening for abnormalities has been recommended. I created a separate missing category due to the high proportion of non-responses.

To measure experiential knowledge, participants reported their experience working in the health field or research field (current; previous; never) and the following relevant histories: number of pregnancies (open), previous pregnancy complications (yes; no; don’t know),

previous prenatal screening (yes; no; don't know), number of children (open), previous NBS (yes; no; don't know), child health history (an acute illness; a chronic illness; a genetic illness; no; do not know), and familiarity with DMD (I know someone diagnosed with DMD; I know someone diagnosed with another genetic disorder; no; do not know).

*Data Analyses*

The primary aim was to examine the influences on intended utilization of optional DMD NBS. To test the main effects of mandatory NBS panel bundling and subjective norms, I used ANOVA to compare the mean intended DMD NBS utilization scores across the experimental groups and Bonferroni post-hoc pairwise comparisons to see which means were different from each other. Then I used ordered logistic regressions to analyze whether bundling, subjective norms, and the interaction of the two predicted choice. I created two regression models: the baseline model included the predictor variables; the secondary regression model added participant demographics. To test whether attitudes towards DMD NBS predicted DMD NBS intent I used ordered logistic regressions. I created two regression models: the baseline regression model included attitudes about the importance of DMD NBS; the secondary model added attitudes about the impact of DMD NBS. These models controlled for age, race, gender, marital/partnered status, previous pregnancy, level of education, and household income. The secondary aim was to test the influences of the study experimental factors on attitudes about DMD NBS and participants' own subjective norms. I used ANOVA and *t*-tests to test the effects of bundling and subjective norms, respectively.

*Sample Size*

The primary outcome variable was DMD NBS utilization, measured by intent to utilize. To calculate sample size I looked at the variance in survey responses observed in the preliminary

dataset. Specifically, I looked at the two most relevant test groups (bundled and unbundled) and compared the mean values (2.53, 2.28) and standard deviations (1.29, 1.29) of the 5-point Likert scale item *worry about the result of DMD NBS*, which was associated with intent to utilize DMD NBS. Based on these numbers and using 80% power and significance of 0.05, N=425 per cell, or a total sample size of N=2,250 was needed.

**Results**

A total of 3,215 surveys were completed. 224 surveys were excluded for participants reporting an age less than 21 per the exclusion criteria, resulting in a final N of Study 1 of 2,991. Participants were predominately white (79.9%), male (52.4%), unmarried/unpartnered (60.1%), and had no children (64.5%). The average age was 29.3 (range 21-82). Participants mostly had a college degree (42.1%) and had a household income of $14,500 to under $35,000 (26.0%). See Table 4 for full participant characteristics.

**Table 4. Study 1 Participant Characteristics (N=2,991)[a]**

| | % (N) |
|---|---|
| *Age (range, mean)* | 21-82, 29.3 |
| 21-35 | 81.1 (2,099) |
| 36 and older | 18.9 (489) |
| *Race* | |
| White | 79.9 (2,387) |
| African American | 5.8 (172) |
| Asian | 8.3 (247) |
| Other | 6.0 (180) |
| *Gender* | |
| Male | 52.4 (1,563) |
| Female | 47.6 (1,419) |
| *Marital Status* | |
| Married/partnered | 39.9 (1,189) |
| Not married/partnered | 60.1 (1,791) |
| *Education Level* | |
| Some high school | 1.1 (34) |
| High school/GED | 8.7 (259) |
| Some college/Tech | 37.1 (1,109) |
| College degree | 42.1 (1,259) |
| Advanced degree | 11.0 (328) |
| *Household Income* | |
| <$14.5k[b] | 15.0 (448) |
| $14.5 to <$35k | 26.0 (776) |
| $35k to <$50k | 19.3 (575) |
| $50k to <$75k | 19.1 (570) |
| $75k - <$100k | 11.1 (330) |
| $100k and over | 9.5 (282) |
| *Previous Pregnancy*[c] | |
| Yes | 36.0 (1,078) |
| No | 63.0 (1,883) |
| Don't Know | 1.0 (30) |
| *Number of Children* | |
| 0 | 64.5 (1,930) |
| 1 | 16.8 (503) |
| 2 or more | 18.7 (558) |

[a]N varies due missing data. Percentage of missing data < 0.50% for all variables except for age (13.41% missing)
[b]k=thousand
[c]Own or partner's pregnancy

The primary aim was to test the influences on intended utilization of optional DMD NBS. First I looked at the effect of bundling the mandatory NBS panels. There was a significant difference across groups in the mean scores for likelihood of choosing DMD NBS (F=5.79, p=0.003, See Table 5), but post-hoc analyses revealed no significant difference between the bundled and unbundled groups. Instead, DMD NBS intent scores for participants in the no panel group were significantly lower than the other two groups. From the regression models, the bundling of the mandatory NBS panel did not influence the DMD NBS decision as hypothesized. However people given information about DMD NBS with either a bundled or unbundled mandatory NBS panel were more likely to choose DMD NBS, compared to those given information without the context of a mandatory NBS panel (OR=1.44, CI=1.10, 1.89, p<0.01; OR=1.34, CI=1.03, 1.74, p<0.05, respectively, see Table 6, Model 1). These findings remained significant after controlling for demographic characteristics (Table 6, Model 2).

**Table 5. Likelihood of Choosing DMD NBS, by Mandatory NBS Panel Bundling and Subjective Norms**

| **Subjective Norms**[a] | **Bundling**[a] | | | |
|---|---|---|---|---|
| | *No Panel* | *Bundled* | *Unbundled* | *Total Mean (SD)*[b] |
| *No* | 3.50 | 3.61 | 3.60 | 3.57 (0.77) |
| *Yes* | 3.46 | 3.53 | 3.58 | 3.53 (0.78) |
| *Total Mean (SD)*[c] | 3.48 (0.81) | 3.57 (0.77) | 3.59 (0.74) | |

[a]Mean (SD); 1=very unlikely, 4=very likely
[b]Difference between means across subjective norms groups t=1.35, p=0.18
[c]Difference between means across panel bundling groups F=5.79, p=0.003

There was no significant difference in the means of DMD NBS intent between the two subjective norms groups (Table 5), nor was choosing DMD NBS predicted by the presentation of additional subjective norms information (OR=0.90, CI=0.70, 1.15, n.s.; See Table 6, Model 1). Although there was a main effect of bundling, there were no interaction effects between mandatory NBS panel bundling and subjective norms. Certain demographic characteristics predicted likelihood of choosing DMD NBS (Table 6, Model 2). Female participants were more

likely than their male counterparts to choose DMD NBS (OR=1.19, CI=1.00, 1.42, $p<0.05$). African American participants (compared to white participants) and participants with a high school degree or less (compared to their more educated counterparts with a college or advanced degree) were less likely to choose DMD NBS (OR=0.63, CI=0.44, 0.89, $p<0.01$; OR=0.75, CI=0.56, 0.99, $p<0.05$, respectively). Pregnancy history played an important role; participants with a previous pregnancy (either their own or their partner's) were less likely to choose DMD NBS (OR=0.65, CI=0.53, 0.80, $p<0.001$).

**Table 6. Study 1 Test Characteristic and Demographic Predictors of Choosing DMD NBS**

| | MODEL 1 (N=2,991)<br>*Baseline Regression* | MODEL 2 (N=2,562)<br>*+Demographics* |
|---|---|---|
| | OR (95% CI) | OR (95% CI) |
| *Bundling* | | |
| No panel | *Reference* | *Reference* |
| Bundled panel | 1.44 (1.10, 1.89)** | 1.54 (1.14, 2.08)** |
| Unbundled panel | 1.34 (1.03, 1.74)* | 1.35 (1.01, 1.81)* |
| *Subjective norm* | | |
| Norms not shown | *Reference* | *Reference* |
| Norms shown | 0.90 (0.70, 1.15) | 0.89 (0.68, 1.18) |
| *Interaction* | | |
| No panel x norm | *Reference* | *Reference* |
| Bundled x norm | 0.89 (0.61, 1.30) | 0.87 (0.57, 1.31) |
| Unbundled x norm | 1.06 (0.73, 1.53) | 1.04 (0.70, 1.56) |
| *Age* | | |
| 21-35 | --- | *Reference* |
| 36 and older | | 0.94 (0.75, 1.19) |
| *Race* | | |
| White | --- | *Reference* |
| African American | | 0.63 (0.44, 0.89)** |
| Asian | | 1.37 (0.98, 1.90) |
| Other | | 1.10 (0.78, 1.57) |
| *Gender* | | |
| Male | --- | *Reference* |
| Female | | 1.19 (1.00, 1.42)* |
| *Marital Status* | | |
| Married/partnered | --- | *Reference* |
| unmarried/partnered | | 1.12 (0.91, 1.36) |
| *Previous Pregnancy* | | |
| No | --- | *Reference* |
| Yes | | 0.65 (0.53, 0.80)*** |
| *Education Level* | | |
| College/Adv degree | --- | *Reference* |
| Some college/Tech | | 1.12 (0.94, 1.35) |
| High school or less | | 0.75 (0.56, 0.99)* |
| *Income* | | |
| <$35k | --- | *Reference* |
| $35k to <$75k | | 1.15 (0.95, 1.40) |
| $75k and over | | 1.13 (0.89, 1.44) |
| *p<0.05, **p<0.01, ***p<0.001 | | |

The likelihood of choosing DMD NBS was also predicted by attitudes about DMD NBS (Table 7). Model 1 explored attitudes about DMD NBS importance; all three were significant predictors. Participants who reported higher importance of DMD NBS were over three times as likely to choose DMD NBS than those who reported lower importance (OR=3.73, CI=3.24, 4.29, $p<0.001$). Similarly, higher reported importance of seeing the DMD NBS results was associated with increased likelihood of choosing DMD NBS (OR=2.36, CI=2.06, 2.70, $p<0.001$). The third "importance" measure, how important it is to share the DMD NBS results with others, was also significant, but participants reporting high importance for sharing results (vs. low importance) were *less* likely to choose DMD NBS (OR=0.87, CI=0.80, 0.94, $p<0.01$). Model 1 controlled for bundling, subjective norms, and the demographic characteristics included in Table 6 Model 2. African American race ($p<0.05$), previous pregnancy ($p<0.001$), high school education or less ($p<0.001$) and a bundled mandatory NBS panel ($p<0.01$) remained significant factors.

The likelihood of choosing DMD NBS was also predicted by two attitudes about the impact of DMD NBS (Table 7, Model 2). Participants who reported that the information from DMD NBS may help prepare for the future were over two times as likely to choose DMD NBS (OR=2.26, CI=1.94, 2.63, $p<0.001$), and participants reporting worry about the DMD NBS results were less likely to choose DMD NBS (OR=0.67, CI=0.60, 0.73, $p<0.001$). The impact of DMD NBS results on reproductive planning, how you might treat your child, and how others might treat your child were not significant predictors. Adding these additional attitude measures did not change the significance of reported importance of DMD NBS (OR=3.56, CI=3.05, 4.15, $p<0.001$) or seeing the DMD NBS results (OR=2.14, CI=1.86, 2.47, $p<0.001$), however, reporting importance of sharing the DMD NBS results was no longer a significant predictor of DMD NBS (OR=0.95, CI=0.87, 1.04, n.s.).

Turning to participants' own subjective norms (Table 7, Model 3), subjective norms around NBS in general and subjective norms around DMD specifically were not equally as important. There was no association between DMD NBS intent and subjective norms around NBS in general, but participants reporting higher subjective norms around DMD NBS specifically were more likely to choose DMD NBS (OR=1.26, CI=1.14, 1.41, $p<0.001$). All three regression models controlled for the main effects, interaction effects, and demographic characteristics in Table 6 Model 2.

**Table 7. Study 1 Attitude Predictors of Choosing DMD NBS**[a]

| | | MODEL 1[b] (N=2,554) *Attitudes about DMD NBS importance* | MODEL 2[c] (N=2,537) *+ Attitudes about the impact of DMD NBS* | MODEL 3[d] (N=2,507) *+ Subjective norms* |
|---|---|---|---|---|
| | Mean (SD) | OR (95% CI) | OR (95% CI) | OR (95% CI) |
| Importance of DMD NBS | 4.07 (1.05) | 3.73 (3.24, 4.29)*** | 3.56 (3.05, 4.15)*** | 3.18 (2.71, 3.72)*** |
| Importance of seeing the DMD NBS results | 4.42 (0.97) | 2.36 (2.06, 2.70)*** | 2.14 (1.86, 2.47)*** | 2.10 (1.82, 2.42)*** |
| Importance of sharing the DMD NBS results | 2.64 (1.33) | 0.87 (0.80, 0.94)** | 0.95 (0.87, 1.04) | 0.91 (0.83, 1.00)* |
| Worry about the DMD NBS results | 3.47 (1.22) | --- | 0.67 (0.60, 0.73)*** | 0.62 (0.56, 0.69)*** |
| The results may help prepare you for the future | 4.40 (0.80) | --- | 2.26 (1.94, 2.63)*** | 2.02 (1.72, 2.36)*** |
| The results may affect if you have more children | 3.20 (1.20) | --- | 0.99 (0.90, 1.09) | 0.98 (0.89, 1.08) |
| The results may affect how you might treat your child | 2.91 (1.34) | --- | 0.96 (0.88, 1.05) | 0.96 (0.88, 1.05) |
| The results may affect how others treat your child | 3.63 (1.02) | --- | 1.08 (0.96, 1.22) | 1.07 (0.95, 1.21) |
| Subjective norms around NBS in general | 5.48 (1.56) | --- | --- | 1.10 (0.99, 1.22) |
| Subjective norms around DMD NBS specifically | 5.55 (1.56) | --- | --- | 1.26 (1.14, 1.41)*** |

[a]Models controlled for all factors in Table 3, Model 2
[b]Significant control factors: African American race ($p<0.05$), previous pregnancy ($p<0.01$), high school education or less ($p<0.001$), bundled panel ($p<0.01$) and unbundled ($p<0.05$) panel
[c]Significant control factors: Age ($p<0.05$), previous pregnancy ($p<0.01$), HS education or less ($p<0.001$), bundled panel ($p<0.01$) and unbundled panel ($p<0.05$)
[d]Significant control factors: Previous pregnancy ($p<0.05$), HS education or less ($p<0.01$), bundled panel ($p<0.01$)
*$p<0.05$, **$p<0.01$, ***$p<0.001$

The secondary aim was to look at the effect of the manipulated study conditions on DMD NBS attitudes. There were effects of mandatory NBS panel bundling (Table 8). Participants who read about DMD NBS without the context of a mandatory NBS panel reported DMD NBS to be less important and reported that they would worry more about DMD NBS results than those who saw either a bundled or unbundled panel ($F=3.40$, $p<0.05$; $F=3.48$, $p<0.05$, respectively). Participants' own subjective norms (i.e. reporting how they think what most other people believe) were associated with bundling; those who viewed DMD NBS without the context of a mandatory NBS panel (either unbundled or bundled) reported weaker norms around the endorsement of both NBS in general and DMD NBS specifically than those viewing either an unbundled or bundled panel ($F=20.01$, $p<0.001$; $F=9.29$, $p<0.001$, respectively).

**Table 8. Attitudes about DMD NBS, by Mandatory NBS Panel Bundling**

| | Panel Bundling | | | |
|---|---|---|---|---|
| | Bundled *Mean (SD)* | Unbundled *Mean (SD)* | No Panel *Mean (SD)* | F |
| Importance of DMD NBS | 4.08 (1.09) | 4.12 (1.00) | 4.00 (1.05) | 3.40* |
| Importance of seeing the DMD NBS results | 4.42 (0.98) | 4.46 (0.90) | 4.38 (1.01) | 1.80 |
| Importance of sharing the DMD NBS results | 2.68 (1.34) | 2.62 (1.31) | 2.61 (1.35) | 0.74 |
| Worry about the DMD NBS results | 3.42 (1.25) | 3.45 (1.20) | 3.56 (1.20) | 3.48* |
| The results may help prepare you for the future | 4.39 (0.83) | 4.41 (0.79) | 4.39 (0.77) | 0.21 |
| The results may affect if you have more children | 3.21 (1.22) | 3.19 (1.19) | 3.20 (1.21) | 0.08 |
| The results may affect how you might treat your child | 2.89 (1.35) | 2.93 (1.34) | 2.92 (1.35) | 0.24 |
| The results may affect how others treat your child | 3.58 (1.06) | 3.64 (0.96) | 3.66 (1.02) | 1.73 |
| Subjective norms around NBS in general | 5.64 (1.52) | 5.71 (1.49) | 5.30 (1.63) | 20.01** |
| Subjective norms around DMD NBS | 5.53 (1.57) | 5.60 (1.54) | 5.31 (1.56) | 9.29** |
| *p<0.05, **p<0.01 | | | | |

There was a group difference in only one attitude measure when looking at the subjective norms study condition (Table 9). Participants who saw additional subjective norm information about how most other parents choose DMD NBS reported that the DMD NBS results would have less of an effect on whether they might have more children than those who did not see additional subjective norm information. ($t=3.10$, $p<0.01$).

**Table 9. Attitudes about DMD NBS, by Subjective Norm**

| | Subjective Norms | | |
|---|---|---|---|
| | Present *Mean (SD)* | Absent *Mean (SD)* | t |
| Importance of DMD NBS | 4.05 (1.05) | 4.08 (1.05) | -0.72 |
| Importance of seeing the DMD NBS results | 4.42 (0.96) | 4.42 (0.98) | -0.20 |
| Importance of sharing the DMD NBS results | 2.62 (1.33) | 2.66 (1.34) | -0.90 |
| Worry about the DMD NBS results | 3.47 (0.03) | 3.47 (0.03) | 0.01 |
| The results may help prepare you for the future | 4.38 (0.80) | 4.42 (0.80) | 1.45 |
| The results may affect if you have more children | 3.14 (1.21) | 3.27 (1.20) | 3.10** |
| The results may affect how you might treat your child | 2.88 (1.35) | 2.94 (1.34) | 1.28 |
| The results may affect how others treat your child | 3.62 (1.00) | 3.63 (1.03) | 0.38 |
| Subjective norms around NBS in general | 5.56 (1.55) | 5.54 (1.56) | -0.24 |
| Subjective norms around DMD NBS | 5.50 (1.54) | 5.46 (1.58) | -0.83 |
| **p<0.01 | | | |

**Discussion**

The primary aim was to test the influences of the study factors on intended utilization of optional DMD NBS. The data did not support my original hypothesis that participants viewing bundled mandatory NBS panels would report higher DMD NBS intention compared to participants seeing unbundled mandatory NBS panels. This null finding was possibly due to a ceiling effect, as the vast majority of participants chose DMD NBS in this study. Despite the null finding, further research on the effects of bundling is needed. For example, the effect of bundling

might be more significant when measured in a clinic during actual decision making, in concert with the stressors of a clinical environment. However I did find that participants given DMD NBS information in the context of either a bundled or unbundled mandatory NBS panel were more likely to choose DMD NBS than those not shown a mandatory NBS panel at all. This finding indicates that when participants were not given the context of broader mandatory NBS in which to place a specific optional NBS, they were more hesitant to choose testing.

Participants viewing additional subjective norm information about parents' DMD NBS utilization did not report higher DMD NBS intention compared to participants not seeing such additional information, contrary to my original hypothesis. However, additional results showed that participants' own subjective norm (what important people in their lives think) towards DMD NBS specifically was a significant predictor of intent. Together these results lend support to TRA; it is the norms of identified people in some one's life that are influential, while the overall norms of unidentified people akin to the participants, whom they do not know, are not influential in decision making. These findings correspond with previous research which has asserted that family members' and friends' experiences and opinions are very influential in NBS decision making (Bradley et al., 1993; Davey et al., 2005). Given the strength of subjective norms when based on specific people the decision maker knows and not a nebulous similar group, developers of health decision materials should be careful not to overemphasize the benefit of language such as "people like you" and instead focus on the important people specific to a decision maker.

The likelihood of choosing DMD NBS was also predicted by attitudes about DMD NBS, following previous research about parents' interest in NBS. Participants reporting higher importance of DMD NBS in general and seeing the DMD NBS results were more likely to choose DMD NBS. However, participants reporting higher importance of sharing the DMD NBS

results were less likely to choose DMD NBS. One possible explanation for this finding is that participants may value the concept of sharing results, but not want to share results that are relevant to them specifically (e.g. their child's NBS results). Instead of facing that cognitive dissonance, they would reject the testing. Similar to previous research, participants reporting that the information from DMD NBS may help prepare for the future were more likely to choose DMD NBS (Parsons et al., 2006; Pelias, 2006), and participants reporting increased worry about the DMD NBS results were less likely to choose DMD NBS (DeLuca, Kearney, Norton & Arnold, 2011; Waisbren et al., 2003). Future NBS programs should address parental worry; it is possible that heightened worry is one element preventing an otherwise desired optional NBS. Unlike previous research (Dhondt, 2010; Parsons et al., 2006), participants' reporting that the information from DMD NBS may help future reproductive choices or lead to discrimination did not predict DMD NBS choice.

Mandatory NBS panel bundling influenced participants' attitudes just as it influenced DMD NBS intention. Participants who read about DMD NBS without the context of mandatory NBS (compared to participants who saw either a bundled or unbundled panel) found DMD NBS less appealing overall. They reported DMD NBS to be less important, that they would worry more about DMD NBS results, and that there were weaker norms around the endorsement of both NBS in general and DMD NBS specifically. Although presenting additional subjective norm information did not affect DMD NBS intent, it did influence reproductive planning. Participants who saw additional information about most parents agreeing to DMD NBS reported that the DMD NBS results would have less of an effect on whether they had more children. This finding is logical; presenting the norm that parents engage in DMD NBS would alleviate the inclination to delay childbirth in order to avoid DMD NBS.

Increased likelihood of DMD NBS intent was predicted by demographic characteristics. Female participants were more likely to choose DMD NBS (vs. male), while participants who were African American (vs. white), had a previous pregnancy (vs. no previous pregnancy), and had a high school degree or less (compared to their more educated counterparts with a college or advanced degree) were less likely to choose DMD NBS. The race/ethnic result may reflect the long history of medical mistrust, especially among racial minorities (Corbie-Smith et al., 2002; Corbie-Smith et al., 1999).

Overall, these results communicate that viewing DMD NBS information without the larger context of mandatory NBS plays an important role in decision making. The provision of this context (regardless of a bundled or unbundled format) affected DMD NBS intent as well as attitudes about DMD NBS. Offering additional subjective norm information about parents akin to the participants did not have an effect, but participants' own subjective norms about people important in their own lives had a very strong one. Future optional NBS programs should be careful to sufficiently present the testing information within an appropriate context while recognizing that decision makers may not relate to the choices of unfamiliar, albeit similar, cases.

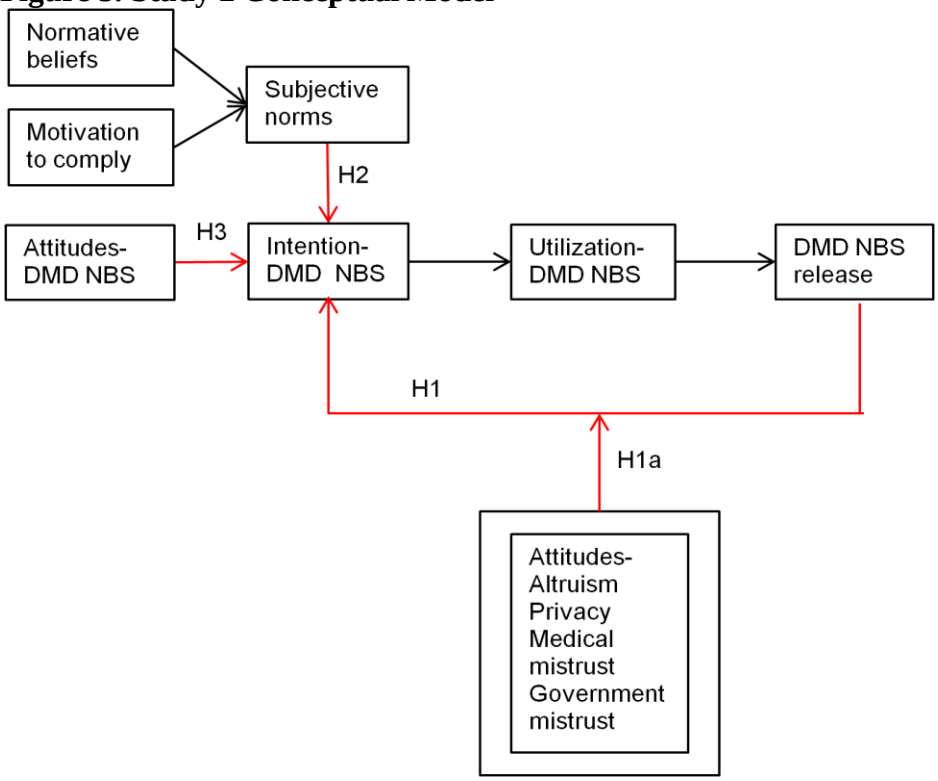
# CHAPTER VI

# STUDY 2

## Introduction

After NBS is complete, the information from the results is released. How, and to whom, this information is released is a test structure feature that can vary from test to test. While previous research has studied parental preferences for the way in which NBS information is released and its effect on psychosocial outcomes, it is unknown whether advance knowledge about how the NBS information will be released effects outcomes of interest. Study 2 considered this possible determinant for the initial decision to utilize DMD NBS.

**Figure 3. Study 2 Conceptual Model**



The conceptual model for Study 2 (Figure 3, above) describes hypothesized influences on DMD NBS intention. In the DMD NBS process, shown in the conceptual model, there are two main action steps: 1. utilizing the test; 2. releasing the results of the test. I hypothesized that when faced with the DMD NBS decision, knowing how the results are to be released would influence the decision-making process; a feedback loop would exist from *DMD NBS release* to the *DMD NBS intent* (H1).

Additionally, the release of DMD NBS results could interact with related personal beliefs, such as preferences for where personal information should be kept and whether the institutions that collect information are trustworthy. Therefore I hypothesized the relationship between *DMD NBS release* and *DMD NBS intent* would be moderated by attitudes towards *altruism, need for privacy, medical mistrust, and government mistrust* (H1a). A high need for privacy, medical mistrust, or government mistrust would mitigate an effect of increased DMD NBS results release on DMD NBS intention, whereas a high value on altruism may strengthen that relationship, as they consider the sharing of DMD NBS results as an altruistic way to contribute to future medical treatments and/or cures. Main effects would parallel these hypotheses: participants with high need for privacy, medical mistrust, or government mistrust would have less DMD NBS intent.

I hypothesized that participant's *subjective norms* associated with NBS in general and DMD NBS specifically would be associated with DMD NBS intention (H2). Reporting more positive subjective norms will be associated with higher likelihood of DMD NBS intent. Finally, I hypothesized that attitudes about DMD NBS would influence DMD NBS intent, with more positive attitudes about DMD NBS associated with higher intent (H3).

*Study Aims and Hypotheses*

Primary Aim: To examine influences of the study experimental factors on intended DMD NBS utilization.

H1: As the DMD NBS results are released to more people in addition to the parents, participants will be less likely to choose DMD NBS.

a) This relationship is moderated by attitudes towards altruism, need for privacy, medical mistrust, and mistrust in government, such that the hypothesized relationship will be mitigated for those with high altruism and attenuated for those with high need for privacy, high medical mistrust, and high government mistrust.

H2: Reporting more positive subjective norms around NBS in general and DMD NBS specifically will be associated with higher DMD NBS intention

H3: Reporting more positive attitudes about DMD NBS will be associated with higher DMD NBS intention

**Study Design**

The study was a randomized survey experiment using a single factor with 5 experimental groups, varying by how the information from DMD NBS is released. Participants were randomly assigned to the experimental conditions:

| | Private | EHR | Research Biobank | Registry: DMD | Registry: General |
|---|---|---|---|---|---|
| **DMD NBS results release** | 1 | 2 | 3 | 4 | 5 |

Release of results is what would happen to the information from the DMD NBS results. Participants were randomized into one of five groups that varied the study brochure's information about the type of results release: private release; EHR; research biobank; DMD

registry; and general registry. Participants in the private release group read that the DMD NBS results would be returned to the family by a private NBS company, and no others would have access to the results unless given permission. Participants in the EHR group read that the DMD NBS results would be viewable to the medical professionals associated with the child, both present and future, in addition to the family having access to the results. Participants in the research biobank condition read that residual blood specimens from the DMD NBS results would be stored in a hospital biobank to be used for future research, in addition to the family having access to the results. There were two groups that were presented with registry scenarios. The first read about a DMD-specific national registry in which the DMD NBS result information would be entered, similar to an existing Duchenne Connect patient registry with over 2,500 registrants from 100 countries (Duchenne Analytics, 2011). This registry would have implications for future DMD treatment and testing options, and therefore individual benefit for those diagnosed with DMD. The second group read about a general national registry that follows overall trends in children's health. The DMD NBS result information would be entered in this registry, but it would have no direct implications for DMD treatment or individual benefits for those enrolled regarding DMD.

## Methods

### *Summary of Methodology*

The methodology for Study 2 is similar to that for Study 1. Briefly, participants above the age of 21, living in the United States and able to complete a web-based survey were paid $0.75 to complete an MTurk-administered Internet study. To begin each study, participants read a vignette that outlined a hypothetical situation in which they have a newborn son and then a brochure that provided a detailed description of DMD, DMD NBS, and DMD NBS results. The

basic information about the hypothetical scenario, DMD and DMD NBS remained the same as in the Study 1 brochures. However, the Study 2 brochure manipulated the information about the release of the DMD NBS results based on the experimental design (see Appendix F for Study 2 vignettes and Appendix G for Study 2 brochures). Then, as in Study 1, participants completed survey items that measured DMD NBS intention, attitudes and beliefs, subjective norms, and demographics (see Study 1 for a detailed methods; see Appendix E for complete survey items).

*Additional Survey Measures for Study 2*

Study 2 focused on the release of results as a predictor of DMD NBS intent. As described in the conceptual model, I hypothesized that this relationship is moderated by beliefs related to how personal information should be used and trust. Four attitudes that capture such beliefs are: altruism, need for privacy, medical mistrust, and government mistrust. Thus participants in Study 2 completed additional survey items to assess these constructs.

I measured altruism using eight "safe" altruism items from the 14-item altruism scale adapted by Homant (2010). I excluded the six "risky" altruism items, which include behavior such as giving a ride to a stranger or making change for a stranger, as they were not relevant to the study. The "safe" altruism items include donating to a charity, volunteering, and simple, everyday courtesies that might indicate a desire to help others through varying degrees of contribution to the larger medical field (see Appendix E for a full description of items). Participants reported how frequently they committed these acts on a 5-point Likert scale from Never to Very Often. A total altruism score was calculated by averaging each item, with higher levels indicating higher altruism.

To measure need for privacy I turned to the 15-item Concern for Information Privacy instrument ([CFIP]; Smith, Milberg & Burke, 1996), which was designed to measure levels of

concern about information privacy practices with respect to data collection, data errors, improper access of data, and unauthorized secondary use of data. I used the 4-item data collection subscale that focuses on sharing personal information, and assigns an overall need for privacy score by averaging each item on 7-point Likert scale from Strongly Disagree to Strongly Agree, with higher levels indicating a higher need for privacy. See Appendix E for a full description of items.

I measured medical mistrust with the 10-item Health Care System Distrust Scale (Rose, Peters, Shea & Armstrong, 2004), which asks participants their agreement with statements about health care system mistakes, testing/experiments without consent, access to medical records, and quality of care (see Appendix E for full description of items) on a 5-point Likert scale from Strongly Disagree to Strongly Agree. An overall medical mistrust score was calculated by averaging each item, with higher levels indicating higher medical mistrust.

To measure government mistrust I used a 6-item validated scale from Peters & Slovic (1996) that measures trust in the government/public officials resolving problems, withholding information, and contributing to problems on a 4-point Likert scale from Strongly Agree to Strongly Disagree (see Appendix E for a full description of items). An overall government mistrust score was calculated by averaging each item, with higher levels indicating higher government mistrust.

*Data Analyses*

The analysis plan followed the same principles and methods as in Study 1. To test the effects of release of DMD NBS results I used ANOVA and Bonferroni post-hoc pairwise comparisons to compare the mean intended DMD NBS intent scores across the experimental groups and ordered logistic regressions to analyze whether results release predicted choice, using private release as the reference group. I created three regression models: the baseline model

included the results release manipulation; the secondary model added altruism, need for privacy, medical mistrust and government mistrust; the tertiary model added participant demographics. I used ordered logistic regressions for each moderating variable to test for interactions as well as main effects. To test whether participants' attitudes towards DMD NBS and their own subjective norms predicted DMD NBS choice I used logistic regressions. I created three models: the first model included attitudes about the importance of DMD NBS; the secondary model added attitudes about the impact of DMD NBS; the tertiary model added participants' subjective norms. All regressions controlled for age, race, gender, marital/partnered status, previous pregnancy, level of education, and household income.

*Sample Size*

My primary outcome variable was intent to utilize DMD NBS. Because the Study 2 factors were not explicitly addressed in the preliminary dataset, I looked at the most relevant factor: reporting the information from DMD NBS as being important information. I used the two groups of people who reported the information being important and not important and compared the mean values and standard deviations of the 5-point Likert scale item *likelihood to choose DMD NBS*. Using 80% power and significance of 0.05, N=310 per cell, or a total sample size of N=1,550 was needed.

**Results**

A total of 1,604 surveys were completed. Similar to Study 1, participants were predominately white (80.5%), male (60.5%), unmarried/unpartnered (66.3%), and had no children (75.8%). The average age was 29.5 (range 21-74). Participants mostly had a college degree (41.0%) and had a household income of $14,500 to under $35,000 (23.5%). See Table 10 for full participant characteristics.

**Table 10. Study 2 Participant Characteristics (N=1,604)[a]**

| | % (N) |
|---|---|
| *Age (range, mean)* | 21-74, 29.5 |
| 21-35 | 80.5 (1,164) |
| 36 and older | 19.5 (282) |
| *Race* | |
| White | 80.5 (1,283) |
| African American | 4.8 (77) |
| Asian | 9.5 (151) |
| Other | 5.2 (82) |
| *Gender* | |
| Male | 60.5 (630) |
| Female | 39.5 (963) |
| *Marital Status* | |
| Married/partnered | 33.7 (539) |
| Not married/partnered | 66.3 (1,059) |
| *Education Level* | |
| Some high school | 0.9 (14) |
| High school/GED | 9.3 (148) |
| Some college/Tech | 38.0 (606) |
| College degree | 41.0 (655) |
| Advanced degree | 10.8 (173) |
| *Income* | |
| <$14.5k | 16.4 (262) |
| $14.5 to <$35k | 23.5 (375) |
| $35k to <$50k | 21.0 (336) |
| $50k to <$75k | 19.1 (305) |
| $75k - <$100k | 9.5 (151) |
| $100k and over | 10.5 (168) |
| *Previous Pregnancy*[b] | |
| Yes | 31.2 (499) |
| No | 67.5 (1,079) |
| Don't Know | 1.3 (20) |
| *Number of Children* | |
| 0 | 75.8 (1,216) |
| 1 | 10.2 (164) |
| 2 or more | 14.0 (224) |

[a]N varies due to missing data. Missing data < 1.0% for all variables except age (10.0%)
[b]Own or partner's pregnancy

Looking at all five groups of results release, I chose to use EHR as the reference group; participants in this group had the highest proportion of DMD NBS intent (Table 11). Although there was no overall statistical significant difference across all five groups (F=1.65, p=0.16, see Table 11), post hoc analyses revealed a significant difference between the EHR release group and the biobank release group ($chi^2$=5.51, p=0.02). Using this reference group, there was a main effect of results release for one comparison (Table 12, Model 1). Participants given DMD NBS with a biobank release were less likely to choose DMD NBS than those given DMD NBS with the EHR release (OR=0.72, CI=0.52, 0.99, p<0.05). This finding remained significant after adding attitudes and participant demographics (Table 12, Models 2 and 3). However no other results release groups were significant.

**Table 11. Likelihood of Choosing DMD NBS, by Mode of Result Release**

| **Results Release**[a] | | | | |
|---|---|---|---|---|
| *Private* | *EHR* | *Biobank* | *DMD Registry* | *General Registry* |
| 3.52 (0.80) | 3.53 (0.81) | 3.39 (0.92) | 3.44 (0.86) | 3.50 (0.85) |
| [a]Mean (SD); 1=very unlikely, 4=very likely<br>Difference between means across results release groups F=1.65, p=0.16 | | | | |

Medical mistrust played a significant role in DMD NBS choice (see Table 12, Model 2). Participants reporting high medical mistrust were significantly less likely to choose DMD NBS than those reporting low mistrust (OR=0.56, CI=0.46, 0.69, p<0.001). Government mistrust was also a significant predictor; overall, participants reporting high government mistrust were significantly more likely to choose DMD NBS (OR=1.43, CI=1.07, 1.90, p<0.05). This result seems counterintuitive, but running a regression without medical mistrust showed an insignificant result (OR=1.02, CI=0.78, 1.32, p=0.91), indicating co-linearity between medical mistrust and government mistrust. Both medical and government mistrust remained significant predictors after controlling for demographic characteristics (Table 12, Model 3).

Similar to Study 1, participants with a previous pregnancy (either their own or their partner's, compared to those without a previous pregnancy) were less likely to choose DMD NBS (OR=0.71, CI=0.53, 0.95, $p<0.05$, Table 12, Model 3). Participants age 36 and older (compared to participants aged 21-35) and with a household income of $75,000 and over (compared to less than $35,000) were also less likely to choose DMD NBS (OR=0.60, CI=0.44, 0.80, $p<0.01$; OR=0.60, CI=0.44, 0.82, $p<0.01$, respectively). As seen in Study 1 there was an effect of race. However, in Study 2 it was self-identifying as Asian that was a significant predictor: Asian participants (vs. white) were more likely to choose DMD NBS (OR=1.60, CI=1.03, 2.48, $p<0.05$).

**Table 12. Study 2 Test Characteristic and Demographic Predictors of Choosing DMD NBS**

| | MODEL 1 (N=1,604) *Baseline Regression* | MODEL 2 (N=1,539) *+Attitudes* | MODEL 3 (N=1,378) *+Demographics* |
|---|---|---|---|
| | OR (95% CI) | OR (95% CI) | OR (95% CI) |
| *Results release* | | | |
| EHR | *Reference* | *Reference* | *Reference* |
| Private | 0.94 (0.68, 1.31) | 0.95 (0.68, 1.33) | 0.97 (0.68, 1.40) |
| Biobank | 0.72 (0.52, 0.99)* | 0.71 (0.51, 0.99)* | 0.69 (0.48, 0.99)* |
| DMD Registry | 0.77 (0.56, 1.06) | 0.75 (0.54, 1.04) | 0.72 (0.50, 1.03) |
| General Registry | 0.93 (0.67, 1.29) | 0.93 (0.67, 1.30) | 0.98 (0.68, 1.40) |
| *Altruism* | --- | 1.04 (0.90, 1.21) | 1.09 (0.93, 1.29) |
| *Need for Privacy* | --- | 1.13 (1.00, 1.28) | 1.14 (0.99, 1.31) |
| *Medical Mistrust* | --- | 0.56 (0.46, 0.69)*** | 0.54 (0.43, 0.67)*** |
| *Government Mistrust* | --- | 1.43 (1.07, 1.90)* | 1.43 (1.04, 1.95)* |
| *Age* | | | |
| 21 - 35 | --- | --- | *Reference* |
| 36 and older | | | 0.60 (0.44, 0.80)** |
| *Race* | | | |
| White | --- | --- | *Reference* |
| African American | | | 0.92 (0.56, 1.50) |
| Asian | | | 1.60 (1.03, 2.48)* |
| Other | | | 0.87 (0.52, 1.46) |
| *Gender* | | | |
| Male | --- | --- | *Reference* |
| Female | | | 1.17 (0.92, 1.50) |
| *Marital Status* | | | |
| Married/partnered | --- | --- | *Reference* |
| unmarried/partnered | | | 0.99 (0.75, 1.30) |
| *Previous Pregnancy* | | | |
| No | --- | --- | *Reference* |
| Yes | | | 0.71 (0.53, 0.95)* |
| *Education Level* | | | |
| College/Adv degree | --- | --- | *Reference* |
| Some college/Tech | | | 1.08 (0.84, 1.39) |
| High school or less | | | 0.89 (0.61, 1.30) |
| *Income* | | | |
| <$35k | --- | --- | *Reference* |
| $35k to <$75k | | | 0.79 (0.61, 1.03) |
| $75k and over | | | 0.60 (0.44, 0.82)** |
| *p<0.05, **p<0.01, ***p<0.001 | | | |

Tables 13a-13d show the interactions of the moderating attitude variables and results release. The interaction model of altruism and results release presents significant findings showing how the effect of altruism was different for the different study groups (Table 13a). First,

there was a main effect of results release. When altruism was not a present characteristic, participants given DMD NBS with a private, biobank, DMD registry or general registry release were all less likely to choose DMD NBS than participants in the EHR results release group (private: OR=0.07, CI=0.01, 0.42, $p<0.01$; biobank: OR=0.09, CI=0.02, 0.47, $p<0.01$; DMD registry: OR=0.10, CI=0.02, 0.55, $p<0.01$; general registry: OR=0.16, CI=0.03, 0.94, $p<0.05$). Second, for only those participants in the EHR results release group, as their altruism scores increased they were less likely to choose DMD NBS (OR=0.67, CI=0.46, 0.99, $p<0.05$). And third, a robust interaction was found. There were changes to the main effect of altruism for each of the private, biobank, DMD registry, and general registry results release groups, with an increase in DMD NBS intent (private: OR=2.17, CI=1.30, 3.63, $p<0.01$; biobank: OR=1.88, CI=1.13, 3.14, $p<0.05$; DMD registry: OR=1.83, CI=1.10, 3.03, $p<0.05$; general registry: OR=1.69, CI=1.01, 2.84, $p<0.05$). So for participants in the EHR group, as altruism increased, intent decreased; for the other participants, as altruism scores increased there was an increase in DMD NBS intent. This was especially the case for those given privately-released DMD NBS. Similar to Table 12, need for privacy was not significant in the interaction model (Table 13b).

Unpacking the main effect of medical mistrust shown in Table 12, the interaction model revealed that for those given DMD NBS with EHR results release, as medical mistrust increased participants were less likely to choose DMD NBS (OR=0.54, CI=0.35, 0.82, $p<0.01$; Table 13c). The interaction model of results release and government mistrust (Table 13d) presents an effect of the biobank results release group. When government mistrust was nonexistent, participants given a biobank results release were less likely to choose DMD NBS than those assigned to the EHR results release (OR=0.07, CI=0.01, 0.81, $p<0.05$). Age over 35, previous pregnancy, and income over $75,000 predicted being less likely to choose DMD NBS in all interactions.

**Table 13a. Study 2 Predictors of Choosing DMD NBS, Interaction of Results Release and Altruism (N=1,421)**[a]

| | OR (95% CI) |
|---|---|
| *Results release* | |
| EHR | *Reference* |
| Private | 0.07 (0.01, 0.42)** |
| Biobank | 0.09 (0.02, 0.47)** |
| DMD Registry | 0.10 (0.02, 0.55)** |
| General Registry | 0.16 (0.03, 0.94)* |
| *Altruism* | 0.67 (0.46, 0.99)* |
| *Interaction* | |
| EHR x Altruism | *Reference* |
| Private release x Altruism | 2.17 (1.30, 3.63)** |
| Biobank x Altruism | 1.88 (1.13, 3.14)* |
| DMD Registry x Altruism | 1.83 (1.10, 3.03)* |
| General Registry xAltruism | 1.69 (1.01, 2.84)* |
| [a]Controlled for demographics variables included in Table 12 Model 3.<br>Significant control factors: Age 36+($p<0.01$), African American race($p<0.05$), previous pregnancy($p<0.01$), household income $75k+ ($p<0.01$)<br>*$p<0.05$, **$p<0.01$ | |

**Table 13b. Study 2 Predictors of Choosing DMD NBS, Interaction of Results Release and Need for Privacy (N=1,420)**[a]

| | OR (95% CI) |
|---|---|
| *Results release* | |
| EHR | *Reference* |
| Private | 0.28 (0.02, 3.13) |
| Biobank | 0.70 (0.07, 6.92) |
| DMD Registry | 0.50 (0.04, 5.57) |
| General Registry | 1.50 (0.12, 18.88) |
| *Need for Privacy* | 1.11 (1.02, 1.46) |
| *Interaction* | |
| EHR x Privacy | *Reference* |
| Private release x Privacy | 1.22 (0.82, 1.82) |
| Biobank x Privacy | 1.00 (0.69, 1.45) |
| DMD Registry x Privacy | 1.06 (0.72, 1.58) |
| General Registry x Privacy | 0.93 (0.62, 1.40) |
| [a]Controlled for demographics variables included in Table 12 Model 3.<br>Significant control factors: Age 36+($p<0.01$), African American race($p<0.05$), previous pregnancy($p<0.05$), household income $75k+ ($p<0.01$)<br>*$p<0.05$ | |

**Table 13c. Study 2 Predictors of Choosing DMD NBS, Interaction of Results Release and Medical Mistrust (N=1,415)[a]**

| | OR (95% CI) |
|---|---|
| *Results release* | |
| EHR | *Reference* |
| Private | 0.73 (0.13, 4.22) |
| Biobank | 0.19 (0.03, 1.15) |
| DMD Registry | 0.42 (0.07, 2.66) |
| General Registry | 1.03 (0.15, 7.19) |
| *Medical Mistrust* | 0.54 (0.35, 0.82)** |
| *Interaction* | |
| EHR x Mistrust | *Reference* |
| Private release x Mistrust | 1.11 (0.62, 1.98) |
| Biobank x Mistrust | 1.56 (0.85, 2.86) |
| DMD Registry x Mistrust | 1.21 (0.66, 2.24) |
| General Registry x Mistrust | 1.00 (0.53, 1.88) |
| [a]Controlled for demographics variables included in Table 12 Model 3.<br>Significant control factors: Age 36+($p<0.01$), previous pregnancy ($p<0.05$), household income $75k+ ($p<0.01$)<br>*$p<0.05$ | |

**Table 13d. Study 2 Predictors of Choosing DMD NBS, Interaction of Results Release and Government Mistrust (N=1,409)[a]**

| | OR (95% CI) |
|---|---|
| *Results release* | |
| EHR | *Reference* |
| Private | 0.84 (0.08, 8.82) |
| Biobank | 0.07 (0.01, 0.81)* |
| DMD Registry | 0.60 (0.05, 6.48) |
| General Registry | 0.24 (0.02, 2.44) |
| *Government Mistrust* | 0.79 (0.43, 1.45) |
| *Interaction* | |
| EHR x Mistrust | *Reference* |
| Private release x Mistrust | 1.05 (0.44, 2.50) |
| Biobank x Mistrust | 2.28 (0.87, 5.54) |
| DMD Registry x Mistrust | 1.08 (0.42, 2.60) |
| General Registry x Mistrust | 1.64 (0.66, 3.87) |
| [a]Controlled for demographics variables included in Table 12 Model 3.<br>Significant control factors: Age 36+($p<0.01$), African American race($p<0.05$), previous pregnancy($p<0.05$), household income $75k+ ($p<0.05$)<br>*$p<0.05$ | |

DMD NBS intent was also predicted by participants' attitudes and subjective norms (Table 14). Similar to Study 1, there was an effect of attitudes about the importance of DMD NBS (Table 14, Model 1). Participants reporting higher importance of DMD NBS were considerably more likely to choose DMD NBS (OR=3.46, CI=2.86, 4.20, $p<0.001$), and participants reporting higher importance of seeing the DMD NBS results were more likely to choose DMD NBS (OR=1.59, CI=1.31, 1.92, $p<0.001$). These results held when considering participants' attitudes about the impact of DMD NBS, which showed to be significant predictors as well (Table 14, Model 2).

Participants reporting that the information from DMD NBS may help prepare for the future higher were more likely to choose DMD NBS (OR=1.81, CI=1.46, 2.24, $p<0.001$) and participants reporting worry about the DMD NBS results were less likely to choose DMD NBS (OR=0.84, CI=0.75, 0.95, $p<0.01$), consistent with Study 1 results. Additionally, participants reporting that the DMD NBS results may affect how they might treat their child were less likely to choose DMD NBS (OR=0.88, CI=0.79, 0.99, $p<0.05$).

The influence of participants' own subjective norms on DMD NBS intent was mixed (Table 14, Model 3). Participants reporting higher subjective norms around NBS in general had no difference in DMD NBS intent than those reporting lower subjective norms. However, participants reporting higher subjective norms around DMD NBS specifically were more likely to choose DMD NBS (OR=1.34, CI=1.16, 1.54, $p<0.001$).

**Table 14. Study 2 Attitude Predictors of Choosing DMD NBS**[a]

| | | MODEL 1[b] (N=1,373) *Attitudes about DMD NBS importance* | MODEL 2[c] (N=1,364) *+ Attitudes about the impact of DMD NBS* | MODEL 3[d] (N=1,347) *+ Subjective norms* |
|---|---|---|---|---|
| | Mean (SD) | OR (95% CI) | OR (95% CI) | OR (95% CI) |
| Importance of DMD NBS | 4.11 (1.00) | 3.46 (2.86, 4.20)*** | 3.12 (2.55, 3.81)*** | 2.86 (2.32, 3.52)*** |
| Importance of seeing the DMD NBS results | 4.45 (0.93) | 1.59 (1.31, 1.92)*** | 1.51 (1.24, 1.83)*** | 1.53 (1.25, 1.86)*** |
| Importance of sharing the DMD NBS results | 2.64 (1.30) | 0.95 (0.84, 1.06) | 0.97 (0.86, 1.09) | 0.95 (0.85, 1.08) |
| Worry about the DMD NBS results | 3.43 (1.19) | --- | 0.84 (0.75, 0.95)** | 0.83 (0.74, 0.93)** |
| The results may help prepare you for the future | 4.40 (0.76) | --- | 1.81 (1.46, 2.24)*** | 1.63 (1.31, 2.03)*** |
| The results may affect if you have more children | 3.17 (1.18) | --- | 0.91 (0.81, 1.02) | 0.91 (0.80, 1.02) |
| The results may affect how you might treat your child | 2.92 (1.33) | --- | 0.88 (0.79, 0.99)* | 0.87 (0.78, 0.98)* |
| The results may affect how others treat your child | 3.35 (1.12) | --- | 1.10 (0.96, 1.25) | 1.10 (0.97, 1.26) |
| Subjective norms around NBS in general | 5.51 (1.46) | --- | --- | 0.97 (0.84, 1.11) |
| Subjective norms around DMD NBS specifically | 5.40 (1.49) | --- | --- | 1.34 (1.16, 1.54)*** |

[a] Models controlled for all factors in Table 12, Model 3.
[b] Significant control factors: Medical mistrust ($p<0.01$), government mistrust ($p<0.05$), income 75K+ ($p<0.01$), previous pregnancy ($p<0.05$)
[c] Significant control factors: Need for privacy ($p<0.05$), medical mistrust ($p<0.05$), income 75K+ ($p<0.01$), previous pregnancy ($p<0.05$)
[d] Significant control factors: Need for privacy ($p<0.05$), medical mistrust ($p<0.05$), income 75K+ ($p<0.01$), previous pregnancy ($p<0.05$)
*$p<0.05$, **$p<0.01$, ***$p<0.001$

## Discussion

The primary aim of Study 2 was to test the influences of the mode of results release on intended utilization of optional DMD NBS. Looking at the influence of the way in which the DMD NBS results would be released, participants in the biobank release group were less likely to choose DMD NBS than those in the EHR release group. However, there were no significant differences between the other modes of results release. These null findings could have occurred for two possible reasons: First, it is possible that the descriptions of the five modes (private release, EHR, biobank, DMD registry, and general registry) had too little variation in the characteristics that would either motivate or discourage utilization. Alternately, it is possible that people do care about mode of release but did not notice the variations across conditions because the study materials did not highlight them. I explore this explanation to some degree in Study 3.

The interaction model of altruism and results release presents significant findings. Focusing on participants given the EHR results release, participants were less likely to choose DMD NBS as they reported increased altruism. The individual characteristic of altruism did not motivate DMD NBS intent for participants in the EHR release group, because that avenue of release had no societal implications. For the non-altruistic participants only, those in the private, biobank, DMD registry or general registry release groups were all less likely to choose DMD NBS than those in the EHR release group. Without the motivation of altruism participants did not choose DMD NBS when its results release would have larger societal contributions. The interaction effect confirmed these two main effects. Comparing all of the other results release groups, as participants' were more altruistic they were more likely to choose DMD NBS. Overall, the findings related to altruism paint a picture about the appropriateness of tapping into

a participants' such motives. Highly altruistic people are not guided by altruism to consent to a testing process if they know that they will not be giving a specific contribution by doing so.

Additionally, the role of participants' medical mistrust was significant; those reporting high medical mistrust were significantly less likely to choose DMD NBS than those reporting low mistrust. This result makes sense; participants who mistrust the institution involved in NBS would not want to engage in DMD NBS and previous research has shown trust in the medical community to be a central attitude to mothers making NBS decisions (Parsons et al., 2006). Such results should be taken into account when informing decision makers about NBS, or any medical procedure in general. The main effect for government mistrust was significant; participants reporting high government mistrust were significantly more likely to choose DMD NBS. Additional interaction analyses showed that when government mistrust was not an issue, participants in the biobank release group were still less likely to choose DMD NBS than those in the EHR release group. This result was unexpected; I hypothesized that it was government mistrust driving the reluctance to choose the DMD NBS with a biobank release. However, this counterintuitive finding might be explained by the measure's colinearity with medical mistrust. There were no main or interaction effects for participants' reported need for privacy.

Increased likelihood of choosing DMD NBS was predicted by higher reported importance of DMD NBS in general and seeing the DMD NBS results. Importance of sharing the DMD NBS did not emerge as a significant predictor of DMD NBS intent. This finding may help explain why I did not see more robust results for the results release variable. Similar to Study 1 participants reporting that the information from DMD NBS may help prepare for the future were more likely to choose DMD NBS, and participants reporting increased worry about the DMD NBS results were less likely to choose DMD NBS. A new attitude emerged in Study 2:

participants reporting that the DMD NBS results may affect how they might treat the child were less likely to choose DMD NBS. This finding is consistent with prior research that DMD NBS may lead to a disruption of the parent/newborn relationship (Bailey et al., 2009; Goddard & Cardinal, 2004). Although subjective norms around genetic testing in general did not predict DMD NBS intent, subjective norms around DMD NBS specifically did. Thus it is possible that in the context of optional testing, it is others' opinions about the specific disease/test that matters.

Similar to Study 1, having a previous pregnancy predicted lower likelihood of DMD NBS intent. Again I saw an effect of race, but it was self-identifying as Asian that predicted higher likelihood of DMD NBS intent. Participants with a high household income and participants over age 35 were less likely to choose DMD NBS than their counterparts, despite the general suggestion that pregnant women over 35 obtain increased genetic testing.

Overall, these results tell a story of participants' hesitancy to choose DMD NBS when the results would be released into a biobank, versus an electronic health record. Participants with low altruism or low government mistrust particularly followed this trend. For those assigned to an EHR release, high medical mistrust and high altruism predicted lower likelihood of choosing DMD NBS. Additionally, a set of significant attitude predictors emerged, consistent with Study 1 and previous literature. Future DMD NBS pilot studies might address medical mistrust as a personal characteristic that biases decision making, and align decision makers' altruistic goals with their ideal mode of results release. Although Study 2 did find robust results with the moderating variables, only two study groups were significantly different. These two groups stood at the far ends of a spectrum: EHR release represented testing with a truly individual goal, and biobank release represented testing with a larger goal to benefit research. These two distinctions were further explored in Study 3.

# CHAPTER VII

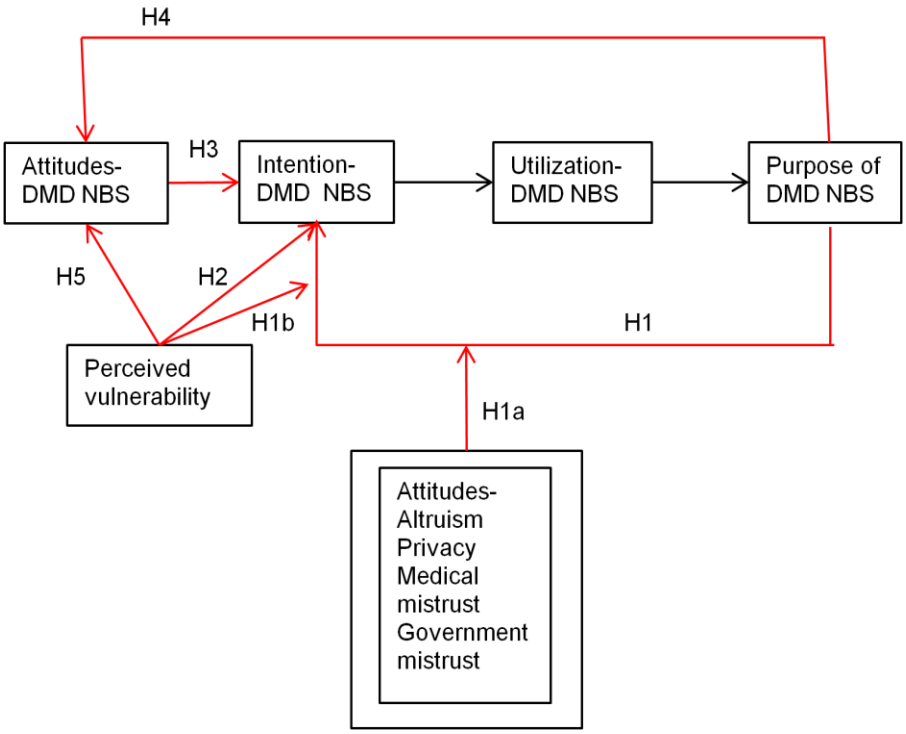
## Study 3

### Introduction

The results from Study 2 showed an effect of only 1 type of results release, a biobank release compared to an electronic health records release. After looking at these results I noted that the groups from Study 2 were nuanced with too little variation in release characteristics between them, especially considering the 2 registry groups. Therefore I developed a third study to look at the theoretical extremes of results release to explore the greatest variation possible, focusing on the driving purpose behind each release type instead of each possible deviation.

Study 3 examined two possible influences on DMD NBS decision making. First, I examined how the purpose for which the DMD NBS results are released may influence the initial decision to utilize DMD NBS. I did not emphasize the specific way that DMD NBS results are released, but instead the overall purpose of the testing that guided the release. Second, I investigated the perceived vulnerability of the child to receive a positive DMD NBS test result, varying from no history to non-genetic health problems at birth.

**Figure 4. Study 3 Conceptual Model**




The conceptual model for Study 3 (Figure 4, above) describes hypothesized influences on the intent to utilize DMD NBS. The model is drawn from the model in Study 2; there are two main action steps in the DMD NBS process: 1. utilizing the NBS; 2. releasing the NBS results. However in Study 3 I focused on the overall *purpose of DMD NBS* and hypothesized that it influences *DMD NBS intention* (H1), as opposed to the specific avenues of release. The test purpose is an important construct; previous research has shown that parents requesting information look for information about the nature and purpose of NBS and not the technical details (Campbell & Ross 2004). Just as in Study2, I hypothesized that this relationship would be moderated by a person' attitudes towards the following: *altruism, need for privacy, medical mistrust, and government mistrust* (H1a).

I hypothesized that participants who read a scenario in which their child was vulnerable to DMD (i.e. had a DMD family history) would be more likely to choose DMD NBS than those

with a no history scenario (H2). This effect of *perceived vulnerability* would also be seen in the other conditions; I hypothesized that there would be a difference in DMD NBS intent between the no history condition and the epilepsy history and premature birth conditions. This construct perceived vulnerability would also interact with test purpose (H1b). Similar to both Study 1 and 2, I hypothesized that *attitudes towards DMD NBS* would predict DMD NBS intent (H3). In addition to predictors of DMD NBS intent, Study 3 addressed influences on attitudes towards DMD NBS. I assessed the association between attitudes and the two study factors, test purpose (H4) and perceived vulnerability (H5).

*Study Aims and Hypotheses*

Primary Aim: To examine the influences on intended utilization of DMD NBS.

H1: Participants presented with DMD NBS for research purposes will report lower DMD NBS intention, compared to participants presented with DMD NBS for personal purposes

a) This relationship is moderated by attitudes towards altruism, need for privacy, medical mistrust, and mistrust in government, such that the hypothesized relationship will be mitigated for those with high altruism and attenuated for those with high need for privacy, high medical mistrust, and high government mistrust.
b) This relationship is moderated by participants' perceived vulnerability as manipulated in the scenario

H2: Participants presented with a high vulnerability scenario will report higher DMD NBS intention, compared to participants presented with no DMD NBS history

H3: Reporting more positive attitudes about DMD NBS will be associated with higher DMD NBS intention

Secondary Aim: To examine the influences of the experimental factors on attitudes about optional DMD NBS

H4: Participants presented with DMD NBS for personal purposes will report more positive attitudes about DMD NBS, compared to participants presented with DMD NBS for research purposes

H5: Participants with more relevant perceived vulnerability will report more positive attitudes about DMD NBS

**Study Design**

Study 3 had a 4x2 between-subjects experimental design: perceived vulnerability (no family history of DMD, family history of DMD, family history of another neurological disease, premature birth) x test purpose (personal, research). Participants were randomly assigned to one of the eight experimental groups:

| | | **Perceived Vulnerability** | | | |
|---|---|---|---|---|---|
| | | No Family History | Family history of DMD | Family history of another | Premature birth |
| **Test Purpose** | Personal | 1 | 2 | 3 | 4 |
| | Research | 5 | 6 | 7 | 8 |

Perceived vulnerability describes how vulnerable the decision makers think the child is for DMD. Participants were randomized to one of the four groups that varied by the level of perceived vulnerability, based on a manipulation of the study scenario: for the no family history group, the scenario described a situation in which no family history of DMD was specified or implicated. The family history of DMD group was just that: the scenario specified that there was a family history of DMD and the doctor wanted to have a further conversation about this history. The participant had already read about DMD so he/she knew that DMD has a genetic component. Participants assigned to the family history of another neurological disease group

read a scenario in which the doctor wanted to have a conversation about a family history of Epilepsy, which was described as a neurological disorder that causes seizures. The purpose of this group was to test whether participants associated the vulnerability of one disorder that affects the brain with the vulnerability of another disorder that affects the brain, despite the massive differences between the two in etiology and health outcomes. The final group read a scenario in which the child was born prematurely and required medical attention at birth, and would require continual medical attention in the future. The purpose of this group was to test whether participant associated a child's overall medical vulnerability with the vulnerability of a specific disorder with a genetic component.

The test purpose described the overall purpose of the DMD NBS that guided what would happen to the information from the DMD NBS results. Participants were randomly assigned to one of two groups based on the study design and the study materials: personal; and research. Participants in the personal purpose group read about DMD NBS done for their family benefit only. For this group, the results would not released to anyone not associated with the family for larger societal goals; in this way the group was theoretically similar to the Study 2 private and EHR release groups. Participants in the research group read about DMD NBS with the societal aims of improving future treatment and testing options for DMD. The results would be viewable not only to the family, but also to unrelated others in order to reach this goal. This group took a broader, more theoretical approach to the Study 2 biobank, DMD registry, and general registry release groups.

## Methods

### *Summary of Methodology*

The methodology for Study 3 was identical to that of Study 2. Briefly, participants above the age of 21, living in the United States and able to complete a web-based survey were paid $0.75 to complete an MTurk-administered Internet study. To begin each study, participants read a vignette that outlined a hypothetical situation in which they have a newborn son and then a brochure that provided a detailed description of DMD, DMD NBS, and DMD NBS results (See Appendix H for Study 3 vignettes and Appendix I for Study 3 brochures). The vignettes differed by how the vulnerability of child to DMD was presented in the scenario, following the study design. The study brochures differed by their description of the DMD NBS. Half presented the overall purpose of DMD NBS as a personal one, and half presented the purpose as a larger societal one to advance research. Then participants completed survey items that measured DMD NBS intention, attitudes and beliefs, subjective norms, attitudes towards altruism, need for privacy, medical mistrust, government mistrust, and demographics (described in Study 1 and Study 2 chapters; see Appendix E for full items). Sample size calculations were identical as for Study 2; a sample size of N=310 per cell, or N=2,480 total was needed for Study 3.

### *Data Analyses*

To test the main effects of perceived vulnerability and test purpose, I used ANOVA to compare the mean across the experimental groups of intended DMD NBS utilization scores and bonferroni post-hoc pairwise comparisons to see which means were different from each other. Then I used ordered logistic regressions to test whether perceived vulnerability, test purpose, and the interaction of the two predicted DMD NBS choice. I used logistic regressions to test for main effects of altruism, need for privacy, medical mistrust, and government mistrust on DMD NBS

choice, as well as interactions with test purpose. To test whether attitudes towards DMD NBS predicted DMD NBS choice I created two logistic regression models: the first included attitudes about the importance of DMD NBS; the second added attitudes about the impact of DMD NBS. All regressions controlled for age, race, gender, marital/partnered status, previous pregnancy, level of education, and household income.

**Results**

A total of 3,090 surveys were completed. Participants were predominately white (80.9%), female (52.0%), unmarried/unpartnered (53.7%), and had no children (65.2%). The average age was 31.6 (range 21-82). Participants mostly had a college degree (42.4%) and had a household income of $14,500 to under $35,000 (26.7%). See Table 15 for full participant characteristics.

**Table 15. Study 3 Participant Characteristics (N=3,090)[a]**

| | % (N) |
|---|---|
| *Age (range, mean)* | 21-82, 31.6 |
| 21-35 | 73.4 (2,095) |
| 36 and older | 26.7 (761) |
| *Race* | |
| White | 80.9 (2,474) |
| African American | 6.9 (207) |
| Asian | 6.8 (212) |
| Other | 5.4 (164) |
| *Gender* | |
| Male | 48.0 (1,471) |
| Female | 52.0 (1, 591) |
| *Marital Status* | |
| Married/partnered | 46.3 (1,419) |
| Not married/partnered | 53.7 (1,648) |
| *Education Level* | |
| Some high school | 1.3 (41) |
| High school/GED | 10.3 (315) |
| Some college/Tech | 34.1 (1,046) |
| College degree | 42.4 (1,300) |
| Advanced degree | 12.0 (367) |
| *Income* | |
| <$14.5k | 12.1 (370) |
| $14.5 to <$35k | 26.7 (816) |
| $35k to <$50k | 20.7 (634) |
| $50k to <$75k | 20.7 (634) |
| $75k - <$100k | 11.4 (350) |
| $100k and over | 8.5 (260) |
| *Previous Pregnancy*[b] | |
| No | 56.3 (1,731) |
| Yes | 43.2 (1,328) |
| Don't Know | 0.6 (17) |
| *Number of Children* | |
| 0 | 65.2 (2,014) |
| 1 | 12.7 (394) |
| 2 or more | 22.1 (682) |

[a]N varies due missing data. Percentage of missing data < 0.50% for all variables except for age (7.6% missing)
[b]Own or partner's pregnancy

The primary aim was to test the influences on intended optional DMD NBS. I studied the effect of the overall test purpose. There was a significant difference between the personal test purpose group and the research test purpose group's mean scores for likelihood of choosing DMD NBS ($t=3.40$, $p<0.001$, see Table 16). The baseline regression model showed no main effect of test purpose on DMD NBS choice, indicating an interaction effect (Table 17). Looking at varying degrees of child's health that would drive perceived vulnerability, there was a significant difference between the mean scores for likelihood of choosing DMD NBS across the four groups ($F=18.58$, $p<0.001$, see Table 16). Post-hoc analyses revealed participants' DMD NBS intent in the highest perceived vulnerability group (DMD history) was significantly higher than that of participants in all 4 other groups. Additional group differences included the no history group and the premature birth group; participants whose hypothetical newborns were born prematurely had higher DMD NBS intent. There was no group difference in DMD NBS intent between the premature birth group and the neurological history (epilepsy history) group. Using the no disease history as the reference category, there was a main effect of perceived vulnerability (Table 17, Model 1). Participants whose hypothetical child had a family history of DMD were much more likely to choose DMD NBS than participants whose child had no family history (OR=3.07, CI=2.21, 4.26, $p<0.001$). Additionally, compared to participants in the no history scenario, participants whose hypothetical child had either a family history of epilepsy or were born prematurely were more likely to choose DMD NBS (OR=1.43, CI=1.08, 1.90, $p<0.05$; OR=1.57, CI=1.18, 2.08, $p<0.01$, respectively).

An interaction was found between test purpose and perceived vulnerability; there was a change to the main effect of test purpose when participants' scenario included either a history of DMD or a history of epilepsy. Participants in these groups were less likely to choose DMD NBS

when the test was for a research purpose compared to a personal purpose (OR=0.60, CI=0.39, 0.92, p<0.05; OR=0.66, CI=0.45, 0.97, p<0.05, respectively). These results were still significant after including demographic characteristics and attitude factors (Table 17, Models 2 and 3).

**Table 16. Likelihood of Choosing DMD NBS, by Perceived Vulnerability and Test Purpose**

| | **Perceived Vulnerability**[a] | | | | |
|---|---|---|---|---|---|
| **Test Purpose**[a] | *No History* | *DMD History* | *Epilepsy History* | *Premature Birth* | Total Mean (SD)[b] |
| *Personal* | 3.29 (0.94) | 3.68 (0.73) | 3.43 (0.90) | 3.49 (0.83) | 3.47 (0.87) |
| *Research* | 3.28 (0.98) | 3.55 (0.80) | 3.28 (0.94) | 3.36 (0.92) | 3.36 (0.92) |
| Total Mean (SD)[c] | 3.29 (0.96) | 3.61 (0.77) | 3.35 (0.92) | 3.43 (0.88) | |

[a]Mean (SD); 1=very unlikely, 4=very likely
[b]Difference between means across test purpose groups t=3.40, p<0.001
[c]Difference between means across perceived vulnerability groups F=18.58, p<0.001

In addition to the manipulated condition, race predicted the likelihood of choosing DMD NBS (Table 17, Model 2). African American participants (compared to white participants) were less likely to choose DMD NBS (OR=0.54, CI=0.41, 0.73, p<0.001). Adding demographic characteristics did not change the main or interaction effects.

**Table 17. Study 3 Test Characteristic and Demographic Predictors of Choosing DMD NBS**

| | MODEL 1 (N=3,090) *Baseline Regression* | MODEL 2 (N=2,812) *+Demographics* | MODEL 3 (N=2,706) *+Attitudes* |
|---|---|---|---|
| | OR (CI) | OR (CI) | OR (CI) |
| *Test Purpose* | | | |
| Personal | *Reference* | *Reference* | *Reference* |
| Research | 1.01 (0.77, 1.32) | 1.02 (0.77, 1.35) | 1.04 (0.78, 1.40) |
| *Perceived Vulnerability* | | | |
| No History | *Reference* | *Reference* | *Reference* |
| DMD History | 3.07 (2.21, 4.26)*** | 3.04 (2.16, 4.30)*** | 3.02 (2.12, 4.31)*** |
| Epilepsy History | 1.43 (1.08, 1.90)* | 1.44 (1.06, 1.94)* | 1.47 (1.08, 2.00)* |
| Premature Birth | 1.57 (1.18, 2.08)** | 1.60 (1.19, 2.16)** | 1.65 (1.22, 2.25)** |
| *Interaction* | | | |
| No History x Research | *Reference* | *Reference* | *Reference* |
| DMD History x Research | 0.60 (0.39, 0.92)* | 0.62 (0.39, 0.99)* | 0.62 (0.39, 0.99)* |
| Epilepsy History x Research | 0.66 (0.45, 0.97)* | 0.65 (0.43, 0.98)* | 0.63 (0.41, 0.96)* |
| Premature Birth x Research | 0.75 (0.51, 1.10) | 0.73 (0.49, 1.11) | 0.70 (0.46, 1.07) |
| *Age* | | | |
| 21-35 | --- | *Reference* | *Reference* |
| 36 and older | | 0.91 (0.76, 1.10) | 0.90 (0.74, 1.09) |
| *Race* | | | |
| White | --- | *Reference* | *Reference* |
| African American | | 0.54 (0.41, 0.73)*** | 0.58 (0.43, 0.78)*** |
| Asian | | 0.77 (0.58, 1.03) | 0.80 (0.59, 1.07) |
| Other | | 0.88 (0.62, 1.24) | 0.82 (0.57, 1.17) |
| *Gender* | | | |
| Male | --- | *Reference* | *Reference* |
| Female | | 1.10 (0.94, 1.28) | 1.06 (0.90, 1.24) |
| *Marital Status* | | | |
| Married/partnered | --- | *Reference* | *Reference* |
| unmarried/partnered | | 0.93 (0.78, 1.11) | 0.96 (0.80, 1.16) |
| *Previous Pregnancy* | | | |
| No | --- | *Reference* | *Reference* |
| Yes | | 0.89 (0.74, 1.07) | 0.88 (0.73, 1.06) |
| *Education Level* | | | |
| College/Adv degree | --- | *Reference* | *Reference* |
| Some college/Tech | | 0.88 (0.74, 1.04) | 0.90 (0.76, 1.07) |
| High school or less | | 0.88 (0.68, 1.14) | 1.00 (0.77, 1.30) |
| *Income* | | | |
| <$35k | --- | *Reference* | *Reference* |

| | | | |
|---|---|---|---|
| $35k to <$75k<br>$75k and over | | 0.93 (0.78, 1.11)<br>0.90 (0.72, 1.12) | 0.95 (0.80, 1.15)<br>0.93 (0.74, 1.16) |
| *Altruism* | --- | --- | 1.09 (0.97-1.22) |
| *Need for Privacy* | --- | --- | 1.13 (1.04-1.23)** |
| *Medical Mistrust* | --- | --- | 0.61 (0.50-0.74)*** |
| *Government Mistrust* | --- | --- | 1.10 (0.97-1.24) |
| *p<0.05, **p<0.01, ***p<0.001 | | | |

Neither altruism nor government mistrust were significant main predictors of DMD NBS choice (see Table 17, Model 3). However there was a main effect of need for privacy and medical mistrust. Participants with a higher need for privacy were more likely to choose DMD NBS, somewhat counterintuitively (OR=1.13, CI=1.04, 1.23, p<0.01). A main effect of medical mistrust showed that participants with higher medical mistrust were less likely to choose DMD NBS (OR=0.61, CI=0.50, 0.74, p<0.001).

I also assessed the interactions of the moderating attitude variables and DMD NBS choice, controlling for the main and interaction effects as well as demographic characteristics in Table 17 Model 3. Similar to Table 17 Model 3, neither altruism nor government mistrust showed interaction effects (see Tables 18a and 18d). No interaction effects were found for either need for privacy (Table 18b) or medical mistrust (Table 18c). For all of the moderating analyses African American race remained a significant control factor of being less likely to choose DMD NBS, as did the main effects of DMD history, epilepsy history, premature birth, and the epilepsy history/research interaction.

**Table 18a. Study 3 Predictors of Choosing DMD NBS, Interaction of Test Purpose and Altruism (N=2,782)**[a]

| | OR (95% CI) |
|---|---|
| *Test Purpose* | |
| Personal | *Reference* |
| Research | 0.76 (0.37, 1.57) |
| *Altruism* | 1.07 (0.91, 1.25) |
| *Interaction* | |
| Altruism x Purpose | 1.00 (0.81, 1.24) |
| [a]Controlled for all factors in Table 17, Model 2. Significant control factors: DMD history(p<0.001), epilepsy history (p<0.05), premature birth (p<0.01), DMD history/ research interaction (p<0.05), epilepsy history/research interaction (p<0.05), African American race (p<0.001) | |

**Table 18b. Study 3 Predictors of Choosing DMD NBS, Interaction of Test Purpose and Need for Privacy (N=2,794)**[a]

| | OR (95% CI) |
|---|---|
| *Test Purpose* | |
| Personal | *Reference* |
| Research | 1.19 (0.44, 3.23) |
| *Need for Privacy* | 1.15 (1.02, 1.30)* |
| *Interaction* | |
| Privacy x Purpose | 1.07 (0.91, 1.26) |
| [a]Model controlled for all factors in Table 17, Model 2. Significant control factors: DMD history(p<0.001), epilepsy history (p<0.05), premature birth (p<0.01), DMD history/ research interaction (p<0.05), epilepsy history/ research interaction (p<0.05), African American race (p<0.001) *p<0.05 | |

**Table 18c. Study 3 Predictors of Choosing DMD NBS, Interaction of Test Purpose and Medical Mistrust (N=2,776)**[a]

| | OR (95% CI) |
|---|---|
| *Test Purpose* | |
| Personal | *Reference* |
| Research | 0.39 (0.12, 1.25) |
| *Medical Mistrust* | 0.58 (0.44, 0.76)*** |
| *Interaction* | |
| Medical Mistrust x Purpose | 0.81 (0.56, 1.16) |
| [a]Model controlled for all factors in Table 17, Model 2. Significant control factors: DMD history(p<0.001), epilepsy history (p<0.05), premature birth (p<0.01), DMD history/ research interaction (p<0.05), epilepsy history/research interaction (p<0.05), African American race (p<0.001) ***p<0.001 | |

**Table 18d. Study 3 Predictors of Choosing DMD NBS, Interaction of Test Purpose and Government Mistrust (N=2,783)[a]**

| | OR (95% CI) |
|---|---|
| *Test Purpose*<br>Personal<br>Research | <br>*Reference*<br>0.63 (0.36, 1.10) |
| *Government Mistrust* | 1.03 (0.87, 1.23) |
| *Interaction*<br>Government Mistrust x Purpose | 0.92 (0.73, 1.16) |
| [a]Model controlled for all factors in Table 17, Model 2. Significant control factors: DMD history($p<0.001$), epilepsy history ($p<0.05$), premature birth ($p<0.01$), epilepsy history/ research interaction ($p<0.05$), African American race ($p<0.001$) | |

DMD NBS intent was also predicted by participants' attitudes and subjective norms (Table 19). A baseline regression model explored attitudes about DMD NBS importance. Similar to the previous studies, I found an effect of attitudes about the importance of DMD NBS (Table 19, Model 1). Participants reporting higher importance of DMD NBS were more likely to choose DMD NBS (OR=2.67, CI=2.35, 3.03, $p<0.001$), and participants reporting higher importance of seeing the DMD NBS results were more likely to choose DMD NBS (OR=1.89, CI=1.67, 2.13, $p<0.001$).

The likelihood of choosing DMD NBS was also predicted by two attitudes about the impact of DMD NBS (Table 19, Model 2). Participants who reported that the information from DMD NBS may help prepare for the future were more likely to choose DMD NBS (OR=1.80, CI=1.57, 2.06, $p<0.001$), and participants reporting worry about the DMD NBS results were less likely to choose DMD NBS (OR=0.70, CI=0.64, 0.75, $p<0.001$). These results are consistent with previous findings. The impact of DMD NBS results on reproductive planning, how you might treat your child, and how others might treat your child were not significant predictors in

this model. Adding these additional attitude measures did not change the significance of reported importance of DMD NBS.

Participants did not put equal weight on subjective norms around NBS in general and around DMD NBS specifically (Table 19, Model 3). Subjective norms around NBS in general did not predict DMD NBS intent, but participants reporting higher subjective norms around DMD NBS specifically were more likely to choose DMD NBS (OR=1.18, CI=1.07, 1.30, $p<0.01$). All regression models in Table 19 controlled for test purpose, perceived vulnerability, attitude measures and demographic characteristics included in Model 3 of Table 17.

**Table 19. Study 3 Attitude Predictors of Choosing DMD NBS**[a]

| | | MODEL 1[b] (N=2,691) *Attitudes about DMD NBS importance* | MODEL 2[c] (N=2,677) *+ Attitudes about the impact of DMD NBS* | MODEL 3[d] (N=2,648) *+ Subjective norms* |
|---|---|---|---|---|
| | Mean (SD) | OR (95% CI) | OR (95% CI) | OR (95% CI) |
| Importance of DMD NBS | 4.11 (1.00) | 2.67 (2.35, 3.03)*** | 2.64 (2.31, 3.01)*** | 2.36 (2.06, 2.71)*** |
| Importance of seeing the DMD NBS results | 4.45 (0.93) | 1.89 (1.67, 2.13)*** | 1.64 (1.45, 1.86)*** | 1.58 (1.40, 1.80)*** |
| Importance of sharing the DMD NBS results | 2.64 (1.30) | 1.01 (0.94, 1.09) | 1.02 (0.95, 1.11) | 1.01 (0.93, 1.09) |
| Worry about the DMD NBS results | 3.43 (1.19) | --- | 0.70 (0.64, 0.75)*** | 0.69 (0.63, 0.74)*** |
| The results may help prepare you for the future | 4.40 (0.76) | --- | 1.80 (1.57, 2.06)*** | 1.72 (1.50, 1.97)*** |
| The results may affect if you have more children | 3.17 (1.18) | --- | 1.05 (0.97, 1.14) | 1.04 (0.95, 1.13) |
| The results may affect how you might treat your child | 2.92 (1.33) | --- | 0.95 (0.88, 1.03) | 0.94 (0.87, 1.02) |
| The results may affect how others treat your child | 3.35 (1.12) | --- | 1.01 (0.93, 1.11) | 1.02 (0.93, 1.11) |
| Subjective norms around NBS in general | 5.51 (1.46) | --- | --- | 1.06 (0.96, 1.16) |
| Subjective norms around DMD NBS specifically | 5.40 (1.49) | --- | --- | 1.18 (1.07, 1.30)** |

[a]Models controlled for all factors in Table 17, Model 3
[b] Significant control factors: DMD history ($p<0.05$), altruism ($p<0.05$), medical mistrust ($p<0.001$), African American race ($p<0.001$), other race ($p<0.05$), high school education or less ($p<0.01$), some college ($p<0.01$)
[c] Significant control factors: DMD history ($p<0.01$), premature birth ($p<0.05$), altruism ($p<0.05$), privacy ($p<0.05$), medical mistrust ($p<0.01$), African American race ($p<0.05$), HS education or less ($p<0.01$), some college ($p<0.01$)
[d] Significant control factors: DMD history ($p<0.01$), premature birth ($p<0.05$), altruism ($p<0.05$), privacy ($p<0.05$), medical mistrust ($p<0.01$), African American race ($p<0.05$), HS education or less ($p<0.01$), some college ($p<0.01$)
*$p<0.05$, **$p<0.01$, ***$p<0.001$

The secondary aim was to look at the effect of the manipulated study conditions on DMD NBS attitudes. There were robust results when comparing perceived vulnerability groups (Table 20). Participants in the different study groups significantly differed in their report of the importance of DMD NBS. Participants with the highest vulnerability group (the DMD history scenario) reported DMD NBS, seeing the DMD NBS results, and sharing the DMD NBS results most important ($F=55.37$ $p<0.001$, $F=16.48$ $p<0.001$, $F=4.96$, $p<0.001$, respectively). For each of these three importance measures, the order of most to least important went: DMD history, epilepsy history, premature birth, and no history. Participants with the DMD history scenario also reported greatest worry about the DMD NBS results ($F=59.39$, $p<0.001$), how much the results would prepare them for the future ($F=20.01$, $p<0.001$), whether the results would affect whether they had more children ($F=17.98$, $p<0.001$). Interestingly, when asked about whether the results would affect how they might treat the child, DMD history did not come into play; participants in the epilepsy history scenario were significantly higher than the lowest score, premature birth ($F=2.85$, $p<0.05$). There was no significant difference between groups in whether the results would affect how others might treat the child.

Participants' own subjective norms (i.e. reporting how they think what most other people believe) were associated with perceived vulnerability; those in the highest vulnerability scenario reported higher norms around the endorsement of both NBS in general and DMD NBS specifically ($F=6.47$, $p<0.001$; $F=28.12$, $p<0.001$, respectively).

**Table 20. Attitudes about DMD NBS, by Perceived Vulnerability**

| | Perceived Vulnerability | | | | |
|---|---|---|---|---|---|
| | No History *Mean (SD)* | DMD History *Mean (SD)* | Epilepsy History *Mean (SD)* | Premature Birth *Mean (SD)* | F |
| Importance of DMD NBS | 3.74 (1.14) | 4.42 (0.87) | 3.97 (1.05) | 3.95 (1.10) | F=55.37*** |
| Importance of seeing the DMD NBS results | 4.21 (1.14) | 4.57 (0.84) | 4.36 (1.02) | 4.31 (1.04) | F=16.48*** |
| Importance of sharing the DMD NBS results | 2.87 (1.33) | 3.13 (1.35) | 2.98 (1.36) | 2.94 (1.37) | F=4.96*** |
| Worry about the DMD NBS results | 3.14 (1.33) | 3.95 (1.12) | 3.30 (1.26) | 3.52 (1.26) | F=59.39*** |
| The results may help prepare you for the future | 4.22 (0.88) | 4.53 (0.71) | 4.36 (0.76) | 4.36 (0.80) | F=20.02*** |
| The results may affect if you have more children | 3.14 (1.18) | 3.56 (1.11) | 3.30 (1.16) | 3.25 (1.20) | F=17.98*** |
| The results may affect how you might treat your child | 2.83 (1.34) | 2.87 (1.38) | 2.96 (1.33) | 2.77 (1.38) | F=2.85* |
| The results may affect how others treat your child | 3.26 (1.16) | 3.29 (1.21) | 3.23 (1.21) | 3.24 (1.14) | F=0.40 |
| Subjective norms around NBS in general | 5.35 (1.56) | 5.69 (1.51) | 5.53 (1.51) | 5.45 (1.59) | F=6.47*** |
| Subjective norms around DMD NBS | 5.19 (1.65) | 5.91 (1.33) | 5.43 (1.55) | 5.45 (1.55) | F=28.12*** |
| *p<0.05, **p<0.01, ***p<0.001 | | | | | |

There was a group difference in multiple attitude measures between the test purpose groups (Table 21). Participants presented with DMD NBS for personal purposes found DMD NBS to be more important ($t=4.53$, $p<0.001$) and reported higher subjective norms around NBS in general and DMD NBS specifically ($t=3.10$, $p<0.001$; $t=3.82$, $p<0.001$, respectively). However, participants presented with DMD NBS for research purposes reported that sharing DMD NBS results to be more important ($t=-2.60$, $p<0.05$).

**Table 21. Attitudes about DMD NBS, by Test Purpose**

| | Test Purpose | | |
|---|---|---|---|
| | Personal *Mean (SD)* | Research *Mean (SD)* | t |
| Importance of DMD NBS | 4.10 (1.05) | 3.92 (1.10) | t= 4.53*** |
| Importance of seeing the DMD NBS results | 4.38 (1.01) | 4.33 (1.04) | t= 1.37 |
| Importance of sharing the DMD NBS results | 2.91 (1.36) | 3.04 (1.34) | t= -2.60* |
| Worry about the DMD NBS results | 3.49 (1.27) | 3.44 (1.30) | t=1.00 |
| The results may help prepare you for the future | 4.39 (0.79) | 4.34 (0.81) | t=1.88 |
| The results may affect if you have more children | 3.29 (1.17) | 3.32 (1.17) | t=-0.54 |
| The results may affect how you might treat your child | 2.85 (1.36) | 2.87 (1.36) | t=-0.28 |
| The results may affect how others treat your child | 3.26 (1.15) | 3.26 (1.17) | t=0.01 |
| Subjective norms around NBS in general | 5.59 (1.51) | 5.42 (1.58) | t= 3.10*** |
| Subjective norms around DMD NBS | 5.59 (1.49) | 5.38 (1.59) | t= 3.82*** |
| *p<0.05, **p<0.01, ***p<0.001 | | | |

**Discussion**

The primary aim of Study 3 was to test the influences on intended utilization of optional DMD NBS, addressing the Study 2 results by considering the overall DMD NBS purpose as opposed to the nuanced modes of results release. A main effect of perceived vulnerability showed that participants whose hypothetical child had a family history of DMD were much more likely to choose DMD NBS than participants whose child had no DMD family history. However other unrelated family histories and health statuses also influenced uptake of DMD NBS;

participants whose hypothetical newborn had either a family history of epilepsy or a premature birth were more likely to choose DMD NBS than participants whose child had no DMD family history. Logically, these two conditions should not differ in DMD NBS intent from the no family history group. This result speaks to an overall sense of vulnerability; participants have a gist reaction to a genetic vulnerability involving a similar part of the body (epilepsy history) or basic health vulnerability (premature birth) and transfer it to vulnerability for other diseases (e.g. DMD).

The purpose of DMD NBS (research vs. personal) did not predict DMD NBS as a main effect. Although this null finding is consistent with the null finding of Study 2, it does not tell the whole story of the role of test purpose. The Study 3 data show important interactions of test purpose with the perceived vulnerability to DMD when participants' scenario included a history of DMD. Participants whose hypothetical newborn had a history of DMD or a history of epilepsy were less likely to choose DMD NBS when it had a research (vs. personal) purpose. This finding indicates that perceived vulnerability to the disease heightens the perceived value of DMD NBS when the screening focuses on personal (vs. research) benefits. This has implications for both the presentation of screening: emphasizing the personal stakes of research, and how it may help participants individually, appears likely to increase consent among those with a personal history of the disease. The results also showed that participants given a family history of epilepsy responded similarly to those with a history of DMD; this may indicate that people see their child with a gist vulnerability when reacting to the personal stakes of research. A similar, albeit non-significant, trend was observed for participants whose hypothetical newborn was born prematurely.

Neither altruism nor government mistrust provided significant main/interaction effects, the former being inconsistent with previous findings. Tabor et al. (2011) found that one of the five main influences of parents' decisions to participate in NBS research is level of altruism. However participants reporting high medical mistrust were less likely to choose DMD NBS, consistent with previous findings (Parsons et al., 2006). Additionally, participants reporting a high need for privacy were more likely to choose DMD NBS. The lack of interaction effects makes this result difficult to interpret.

Study 3 confirmed the trend seen in Studies 1 and 2 of DMD NBS intent predicted by attitudes about DMD NBS. Participants reporting higher importance of DMD NBS in general and seeing the DMD NBS results were more likely to choose DMD NBS. Congruent with previous work, participants reporting that the information from DMD NBS may help prepare for the future were more likely to choose DMD NBS (Parsons et al., 2006; Pelias, 2006), and participants reporting increased worry about the DMD NBS results were less likely to choose DMD NBS (DeLuca et al., 2011; Waisbren et al., 2003). Similar to Study 2, subjective norms around NBS in general did not predict DMD NBS intent, but participants reporting higher subjective norms around DMD NBS specifically were more likely to choose DMD NBS, indicating that specificity of subjective norms is important.

The secondary aim was to look at the influences on DMD NBS attitudes. Participants given the DMD history scenario had the strongest attitudes about DMD NBS; they reported the highest importance of DMD NBS, seeing the DMD NBS results, and sharing the DMD NBS results. This is a logical finding; with the personal connection to the disease participants would favor NBS. Although participants presented with DMD NBS for personal purposes also found DMD NBS to be more important, participants presented with DMD NBS for research purposes

reported that sharing DMD NBS results to be more important. Participants given the DMD history scenario reported the greatest worry about the DMD NBS results, and the most that the results would prepare them for the future and affect whether they had more children. However a history of DMD was not a significant factor regarding whether the DMD NBS results would affect how participants might treat their hypothetical child; instead, participants given a scenario with a history of epilepsy were more likely to answer affirmatively. Participants' own subjective norms (i.e. reporting how they think what most other people believe) were associated with both study factors; those in the highest vulnerability scenario and participants presented with DMD NBS for personal purposes reported higher norms around the endorsement of both NBS in general and DMD NBS specifically.

Participants who were African American (vs. white) were less likely to choose DMD NBS, echoing findings in the previous dissertation studies and by others (Corbie-Smith et al., 2002; Corbie-Smith et al., 1999; Furr, 2022).

Overall, these results communicate that perceived vulnerability to a disorder should not be overlooked. Not only does vulnerability to the relevant disease (e.g. a history of DMD) influence testing decisions, it also influences attitudes towards the test and can change the way people feel about testing for research vs. personal purposes. Interestingly, it appears that vulnerability can be perceived when there is no history of the relevant disease, but instead other health risks. People may process a vulnerability gist concept, instead of focusing on one etiology versus another, and their interest in DMD NBS appears more driven by that gist perception of the child as vulnerable to disease. In addition, Study 3 showed the value of considering test purpose as the overarching, driving force behind the release of results rather than in terms of the narrower, functional definitions used in Study 2. There was an effect of test purpose, in that

perceived vulnerability heightened participants' perceived value of DMD NBS when primed to consider it for a personal purpose. However when the presentation of DMD NBS focused on a research purpose, this did not hold.

# CHAPTER VIII

# Conclusion

## Summary

The focus of this dissertation is whether variation in presentation and characteristics of a medical test influences utilization. To address this larger question I looked specifically at optional newborn screening (NBS), a choice that parents can face in addition to mandatory NBS. This is an issue worth studying because, while parents are not in a decision-making role regarding mandatory NBS panels, with the continued expansion of NBS parents may be increasingly faced with the decision to utilize additional optional NBS. However there is no consistent policy regarding optional NBS communication, in terms of the information included and the way in which this information is presented (Hargreaves et al., 2005; Loeben et al., 1998). These questions will become more and more important because NBS programs are likely to continue expanding, increasingly including diseases for which limited information about treatment efficacy is available at minimal incremental cost (Carroll & Downs, 2006; Insinga et al., 2002; Schoen et al., 2002).

## Findings

In three separate randomized experimental studies I examined possible influences on optional NBS using Duchenne muscular dystrophy (DMD) as an exemplar. DMD provided an ideal paradigm; despite the development of DMD NBS, no treatment is widely used that can alter the disease course of DMD. Therefore learning one's DMD status does not improve clinical

outcomes. Previous pilot programs have offered DMD NBS as an optional NBS but no states have thus far included it their mandatory NBS panels.

*Study 1*

Study 1 explored the test presentation characteristic of bundling mandatory NBS panels with which optional NBS is presented. DMD NBS is presented in the context of mandatory NBS panels; although it is known that unbundling NBS invitations decreases utilization, as opposed to bundling the optional NBS test offer and the mandatory NBS tests (Moody & Choudhry, 2011), this finding did not extend to mandatory NBS panels. Instead, the Study 1 findings showed that it is whether the context of mandatory NBS is discussed in any way (bundled or unbundled) that influences DMD NBS intent. This finding indicates that when participants were not given the context of broader mandatory NBS in which to place a specific optional NBS, they were more hesitant to choose testing. The results suggest that viewing DMD NBS information without the larger perspective of mandatory NBS tempers DMD NBS intent and attitudes about DMD NBS. Future optional NBS programs should be careful to sufficiently present the testing information in a way that provides decision makers with a context in which to understand how that single NBS test fits into NBS overall.

*Study 2*

Study 2 focused on a post-test influence on test decision making that can vary from test to test: the specific test feature of how the NBS results are released. The study addressed whether advance knowledge about NBS results release and related attitudes affects DMD NBS. Although there was no main effect of the modes of results release (private, electronic health record, biobank for future research, DMD registry, and general registry) on DMD NBS intent, two groups differed: participants given a biobank results release were less likely to choose DMD

NBS than those given an EHR release. However, the relatively small effect sizes observed may be attributed to the degree to which I implemented the experimental condition; I used nuanced group divisions (e.g. DMD registry vs. regular registry) and the participants may not have picked up on the subtle differences between the modes of results release. Therefore I re-examined the overarching concept of test purpose that drives results release in a third study.

However, Study 2 did find that increased medical mistrust was as a significant predictor of low DMD NBS intent. This finding is consistent with previous work showing trust in the medical community was central to the attitudes of the mothers considering NBS (Parsons et al., 2006). It also has implications for future NBS program recruitment, signaling that gaining trust, or at least addressing possible existing mistrust, is an important step that might remove testing barriers. The unexpected finding that increased government mistrust predicted DMD NBS intent, may be explained by the measure's colinearity with medical mistrust.

In addition, Study 2 found a significant interaction of results release and altruism. For the non-altruistic participants, those not in the EHR release group were less likely to choose DMD NBS compared to those in the EHR release group, and participants in the EHR results release were less likely to choose DMD NBS as they reported increased altruism. These participants' altruistic motives had no outlet the EHR condition; there are no philanthropic implications of EHR results release. The interaction showed that comparing all of the other results release groups to EHR, as the more altruistic participants were, the more they were likely to choose DMD NBS. These robust findings have implications for future recruitment in NBS programs. Appealing to possible participants' sense of altruism may only be successful if they know their participation will, in fact, be a contribution.

*Study 3*

Study 3 re-examined the results release construct from Study 2, looking at the theoretical extremes to focus on the overarching purpose of the test that drives the mode of results release. The main purpose of DMD NBS (research vs. personal) did not have an effect on DMD NBS intention in the absence of perceived vulnerability sources (e.g. family history), even though the manipulation was more robust than in Study 2. However, interactions with the perceived vulnerability to DMD showed that participants whose hypothetical newborn had a history of DMD or a history of epilepsy were less likely to choose DMD NBS for research purposes (vs. when testing was for personal purposes). This finding suggests that those with related, even marginally so, health experiences think of NBS purposes differently. Focusing parents' attention on personal benefits to NBS may increase utilization.

The main effect of perceived vulnerability showed that participants whose hypothetical child had a family history of DMD were much more likely to choose DMD NBS than participants whose child had no DMD family history. Interestingly, unrelated family histories and health statuses influenced this choice; participants whose hypothetical newborn had either a family history of epilepsy or a premature birth were more likely to choose DMD NBS than participants whose child had no DMD family history, although this effect was smaller than for having a DMD family history. The Study 3 results suggest a gist reaction to a genetic vulnerability involving a similar part of the body (epilepsy history) or basic health vulnerability (premature birth), that is applied to vulnerability for other diseases (e.g. DMD). Health professionals should attend to patients' perceived vulnerability of a disease when there is clear medical cause, as it influences testing decisions, attitudes towards the test and can change the way people feel about testing for research. However they should also attend to perceived

vulnerability when there are other, unrelated health risks that drive patients to conceive of an overall "vulnerable" state.

*Consistent Findings*

A consistent finding throughout the dissertation was that participants were more likely to choose DMD NBS if they reported higher importance of DMD NBS in general and seeing the DMD NBS results, and thought the results could inform future planning. They were less likely to choose DMD NBS if they had increased worry about the DMD NBS results. These results are consistent with previous research (DeLuca et al., 2011; Goddard, 2004; Parsons et al., 2006; Pelias, 2006; Waisbren et al., 2003). There were influences on these attitudes. In Study 1, participants who saw no mandatory NBS panel (vs. bundled or unbundled) thought DMD NBS was less important and would worry more about the DMD NBS results. In Study 3, participants given the DMD history scenario had the strongest attitudes about DMD NBS; they reported the highest importance of DMD NBS, seeing the DMD NBS results, and sharing the DMD NBS results, the greatest worry about the DMD NBS results, and that the results would prepare them for the future and affect whether they had more children the most.

The subjective norm findings are consistent with previous theory and NBS research. Offering additional subjective norm information about parents akin to the participants did not have an effect, but participants' own subjective norms about people important in their own lives had a very strong one. This finding supports the Theory of Reasoned Action, and mirrors previous research asserting that family members' and friends' experiences and opinions are very influential in NBS decision making (Bradley, 1993; Davey, 2005). Additionally, general subjective norms around NBS did not predict DMD NBS intent, but specific subjective norms around DMD NBS did; this indicates that subjective norms are topic-sensitive.

**Strengths and Limitations**

There were multiple strengths of this dissertation. First, it addressed a timely topic for NBS in general and DMD NBS specifically. Over the past year DMD NBS has come to the forefront of optional NBS, with some researchers wanting it to be included in mandatory NBS panels and others calling for more evidence (American Journal of Medical Genetics, 2012). It is necessary to address the evidence base behind the NBS as well as the factors driving parents' NBS decisions before making this change. Second, the vignettes and brochures were carefully crafted with the assistance of a pediatric decision making researcher (B.A. Tarini) and the Statewide Newborn Screening for Duchenne Muscular Dystrophy pilot program in Ohio (Mendell et al., 2012). Third, the theoretical framework was based on classic health behavior theory and grounded in previous NBS literature. Finally, the quantitative experimental design allowed me to cleanly vary specific influences on the NBS decision-making process with little threat to internal validity. I would not have been able to vary these factors using qualitative data collection.

There are also weaknesses to be found in this dissertation. First, despite the effort made to ensure that study materials mirrored those used in real-world contexts, these Internet-administered studies using hypothetical scenarios may not have evoked the same feelings or decision-making processes that would be present in a true population of DMD NBS decision makers, actual parents. Surveying actual parents with more emotional investment in newborn screening and possibly more knowledge may lead to more robust results, especially with the presence of a baby in the room during the decision. Second, although the focus of this experimentally-designed dissertation was to ensure internal validity, the non-generalizability of MTurk subjects is a threat to external validity. Their mere participation indicates that subjects

have the time and internet access to take an MTurk survey, and research has shown that MTurk subjects tend to be younger than the general public (Berinsky et al., 2011). This younger age may contribute to the significant age findings. Finally, by virtue of the study designs and study population, DMD NBS decision making was conceptualized as a solo process and not one done with partners and/or family members. A more likely scenario is a shared decision that includes at least two people (Epstein, 2013). In such a case the presence of additional decision makers may interact with some of the specific factors studied, emphasizing the influence of subjective norms and mitigating the influence one person's perceived vulnerability.

## Implications

This dissertation addressed the question of whether decisions are influenced by characteristics of test presentation and structure, using DMD NBS as a case study. The implications of the results are broad, ranging from improvements to the Theoretical Framework to suggestions for designing health communications related to NBS to advice for clinical practitioners. Many of these implications likely generalize beyond the field of NBS to any health context that includes optional testing.

### *Implications for the Theoretical Framework*

After collecting, analyzing, and interpreting the study data, it is necessary to re-visit the theoretical framework that informed this dissertation (Appendix A). Overall, this framework held as a way of conceptualizing the DMD NBS process into modifying factors, pre-test, and post-test. The results of the three studies validated many of the constructs in the theoretical framework. Looking at the modifying factors, the results confirmed the influence of demographic characteristics, such as race and age, and personal history, such as previous pregnancy. Turning to pre-test factors, results supported the inclusion of subjective norms when

specific to participants' own norms, and attitudes towards both NBS in general and DMD specifically, as these predicted DMD NBS choice throughout the three studies. The post-test factors were partially confirmed in Studies 2 and 3. Although results release did not have a significant main effect on DMD NBS choice in Study 2, the re-framing of the construct in Study 3 as larger test purpose had significant interactions with perceived vulnerability. Additionally, attitudes of altruism, privacy, medical mistrust and government mistrust were inconsistently significant.

However, the results suggest both changes to the theoretical framework, and directions for a new theoretical model. Although Study 1 showed a significant difference in DMD NBS choice between those who did not see a mandatory NBS panel and those who did (either bundled or unbundled), there was no difference between bundled and unbundled mandatory NBS panels. Thus the theoretical framework would best reflect these results if the bundling construct was modified as 'context', or the presence or absence of the mandatory NBS panel. Additionally, given the increasing number of optional NBS tests, a future theoretical framework might include the bundling of optional NBS offerings (as opposed to mandatory NBS panels). Such a construct would potentially tap into the cognitive-style variables such as decision fatigue. In addition, there might be a possible interaction of bundling with perceived vulnerability of the child: given the Study 3 results indicating a gist sense of perceived vulnerability, with more NBS tests to focus on, perceived vulnerability may increase. The perceived vulnerability factor would exist as a pre-test factor in a modified framework. This construct would be predicted by personal history, as seen in Study 3, and knowledge. A last modification to the theoretical framework would be the use of subjective norms only as they are conceptualized by Theory of Reasoned Action and

supported by the results: as norms specific to the disease and to the person, and not general subjective norms.

*Implications for Health Communication*

It is important to understand the implications of the way in which we communicate information about optional NBS. First, if optional NBS is not presented in the context of mandatory NBS, these results suggest that participants' NBS intention will be hindered and they will have more negative attitudes towards NBS. Second, these results suggest that future optional NBS program materials should be as specific as possible in the presentation of subjective norms. Developers should recognize that decision makers may not relate to the choices of unfamiliar, albeit similar, cases and pause before overemphasizing the benefit of common language such as "people like you". Finally, these results suggest that optional NBS, even if in a research context, should not automatically be presented with a research focus. Such a presentation may not appeal to those with a perceived vulnerability to the disease being tested for. For those with a heightened personal stake in the disease, tests with a focus on personal purpose appear to be valued more.

*Clinical Implications*

In addition to the implications for the way in which materials present optional NBS information, these results have direct implications for the way in which doctors interact with parents making optional NBS decisions. First, when parents are making NBS decisions for their children, clinicians should consider assessing their perceived vulnerability to the specific disease as this may influence their decisions. Clinicians should also consider whether parents might have an overall, gist vulnerability stemming from another disease or health risk that could similarly influence the NBS decision. Once clinicians have an understanding of how vulnerable parents

perceive their child to be, they should have a conversation with the parents about the actual risk, especially if that perceived vulnerability stems from an unrelated disease or health issue. Correcting any misconceptions regarding risk may reduce optional NBS overuse (i.e., use by parents who are choosing testing for reasons unrelated to what the test can actually provide). Second, when recruiting parents for optional NBS programs clinicians should be cautious about appealing to parents' sense of altruism, as this can backfire depending on the test result release and the parents' actual altruism. Finally, careful attention must be paid to those with high medical mistrust. Clinicians should approach those with high medical mistrust knowing that this characteristic makes them less likely to choose NBS, and directly address any concerns of mistrust that may exist.

*Policy Implications*

As optional NBS is expanding it is important to consider the possible influences on decision makers before policies and universal guidelines are set. These results inform three larger policy implications for future optional NBS programs. First, there is a need to develop policies regarding the presentation of optional NBS. Although every state has a mandatory NBS program, there are variations in test offering and test presentation among states. These variations inhibit our ability to assess parental interest reliably and create the possibility of greater or lesser testing uptake simply due to structural differences in communications and program design. There is a need to promote consistency among optional NBS programs, including making sure to frame optional NBS within the context of mandatory NBS. Second, parents whose children have family history of related disease appeared to be less motivated by research-focused programs. This result suggests that optional NBS being conducted for research purposes will nonetheless have to call attention to how the NBS will have personal benefits to parents. And finally, the results

related to perceived vulnerability based on unrelated variables (e.g., other diseases) have implications for NBS policy. Those with unrelated disease histories reacted similarly to optional NBS decision making to those with related histories. As a result, future NBS policy might have to consider the potential for possible overuse of optional NBS based on an overall gist perception of vulnerability that is not clinically related to the conditions for which tests are being offered. For example, there may be a need to require that optional NBS programs for parents clarify how different factors that affect perceived child vulnerability (e.g., prematurity) do, or do not, affect the risk of the disease being tested for.

**Directions for Future Research**

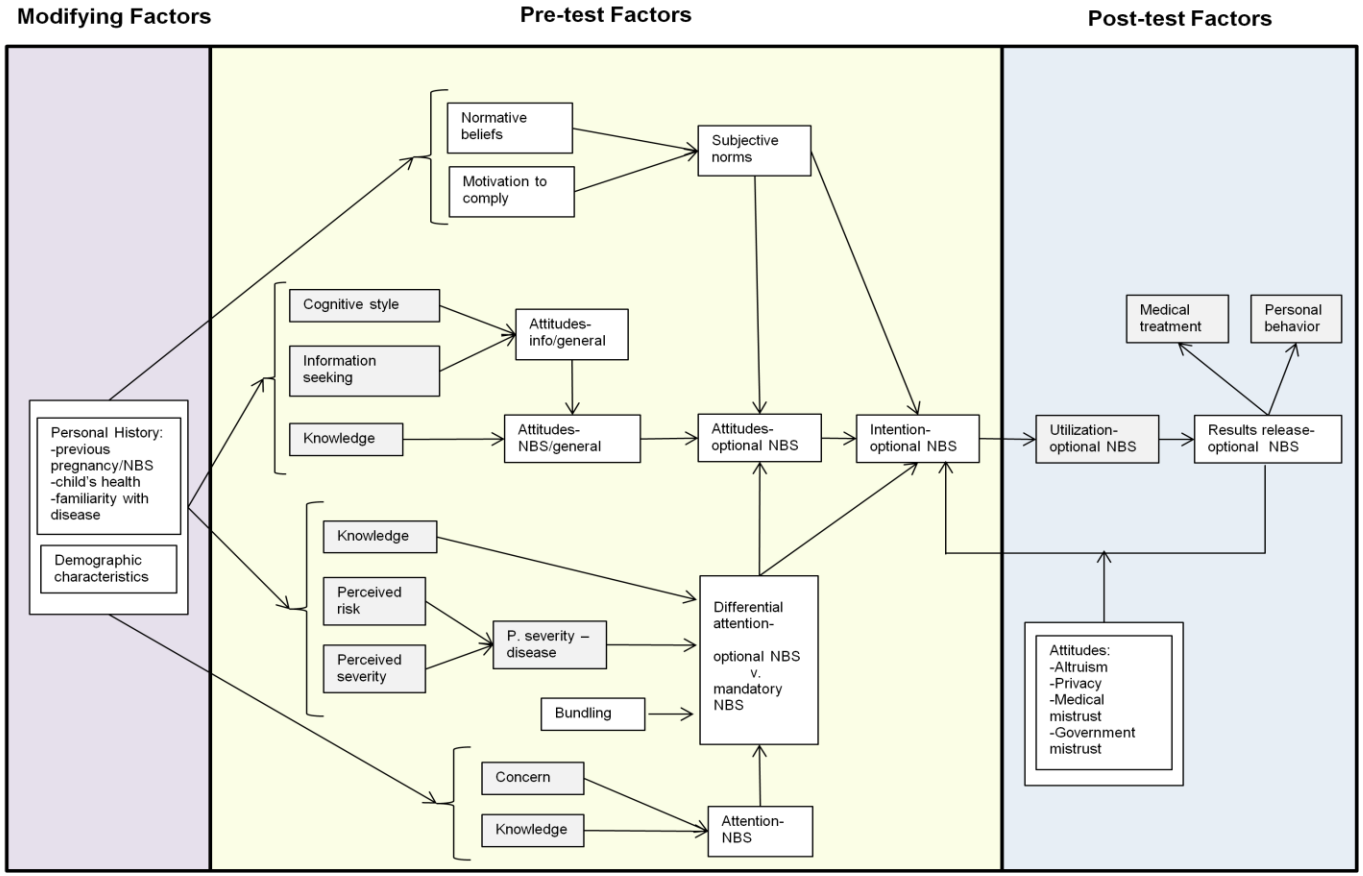
The results of this dissertation have clear implications for future research. Now that we know optional testing is ideally presented in the context of mandatory testing, and given the growth of optional testing, a next step would be to study the bundling of multiple *optional* tests. More research is needed on bundling, and it may be worth looking to the future of multiple optional tests. After seeing the strong effect of *perceived* vulnerability in the study sample, it is a logical to use purposeful sampling of populations that perceive their children to be vulnerable for various reasons. In addition to a main effect, the study would test an interaction of vulnerability and optional test bundling. With the strong subjective norm results that included normative beliefs, further research is needed to identify the important others and address motivation to comply – exactly who are the people that influence decisions, and how does motivation to comply with their norms differ among different types of optional tests?

While technology advances and optional NBS becomes both more available and less expensive, there is a potential for increased optional NBS use. DMD offered an exemplar to study the influences on optional NBS decision making, as knowing how such decisions do vary

by such features may inform future optional NBS practices. But with the push for DMD to become part of the mandatory NBS panel, it will be difficult to study it further as a purely optional NBS test. However, it is not just NBS that is expanding. After the mapping of the genome and the availability of commercialized genetic testing (e.g. 23andMe), the theoretical, clinical, health communication, and policy implications of optional NBS are also relevant for other optional genetic testing. Thus we can study the constructs significant in NBS decision making, like bundling and perceived vulnerability, in other genetic testing contexts. Such study will allow us to understand the influence of test structure and presentation characteristics on patient utilization of genetic tests, and help guide clinical and health communication practice as well as health policy.

# APPENDICES

## Appendix A. Theoretical Framework



## Appendix B. Preliminary Study Vignettes (Experimental Conditions 1-3)

Imagine that you or your partner has just given birth to a baby boy. A nurse comes into your room to tell you that the hospital has developed a new test that screens newborn babies for DMD. DMD cannot be cured and the symptoms cannot be prevented. The test requires a blood sample but causes no long-term harm or side effects. The test will not cost any money.

**[Condition 1 text]** All newborn babies in the state are mandated to be screened for certain diseases. In the hospital after birth he will have a little blood drawn as part of that screening. Using this blood sample, doctors will test for about 49 of these genetic diseases. The sample can also be used to test for Duchenne's. Adding the screening test for Duchenne's will not require you to come to the hospital at an additional time or draw more blood. The results will be automatically released to you. You will need to check a box on the medical chart form.

**[Condition 2 text]** All newborn babies in the state are mandated to be screened for certain diseases. In the hospital after birth he will have a little blood drawn as part of that screening. Using this blood sample, doctors will test the following genetic diseases:

**Amino Acid Disorders**
1. Argininemia
2. Argininosuccinic acidemia
3. Citrullinemia
4. Citrullinemia type II
5. Homocystinuria
6. Hypermethioninemia
7. Maple syrup urine disease (MSUD)
8. Phenylketonuria (PKU)
9. Benign hyperphenylalaninemia defect
10. Biopterin cofactor biosynthesis defect
11. Biopterin cofactor regeneration defect
12. Tyrosinemia type I

**Fatty Acid Oxidation Disorders**
13. Carnitine acylcarnitine translocase deficiency
14. Carnitine palmitoyl transferase I deficiency
15. Carnitine palmitoyl transferase II deficiency
16. Carnitine uptake defect
17. Dienoyl-CoA reductase deficiency
18. Glutaric acidemia type II
19. Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency
20. Medium/short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency
21. Medium-chain acyl-CoA dehydrogenase deficiency
22. Medium-chain ketoacyl-CoAthiolase deficiency
23. Short-chain acyl-CoA dehydrogenase deficiency
24. Trifunctional protein deficiency
25. Very long-chain acyl-CoA dehydrogenase deficiency

**Organic Acid Disorders**
26. 2-Methyl-3-hydroxy butyric aciduria
27. 2- Methylbutyryrl-CoA dehydrogenase deficiency
28. 3-Hydroxy 3-methylglutaric aciduria
29. 3-Methylcrotonyl-CoA carboxylase deficiency
30. 3-Methylglutaconic aciduria
31. Beta-ketothiolase deficiency
32. Glutaric acidemia type I
33. Isobutyryl-CoA dehydrogenase deficiency
34. Isovaleric acidemia
35. Malonic acidemia
36. Methylmalonic academia (Cbl A,B)
37. Methylmalonic academia (Cbl C,D)
38. Methylmalonic acidemia (Mutase deficiency)
39. Multiple carboxylase deficiency
40. Propionic acidemia

**Endocrine Disorder**
41. Congenital adrenal
42. Congenital hypothyroidism (CH) hyperplasia (CAH)

**Hemoglobinopathies**
43. S/Beta thalassemia
44. S/C disease
45. Sickle cell anemia
46. Variant hemoglobinopathies

**Other Disorders**
47. Biotinidase deficiency
48. Cystic Fibrosis
49. Galactosemia

The sample can also be used to test for Duchenne's. Adding the screening test for Duchenne's will not require you to come to the hospital at an additional time or draw more blood. However, because the test is new, even though the results are available they are not automatically released. If you would like to learn the results of the test then you need to fill out a form, have

your pediatrician sign it, and mail it to the hospital laboratory to make a special request after the test has been done and the results are ready.

**[Condition 3 text]** The test for Duchenne's requires a special request, and you will have to bring your child to the hospital at a separate time to have additional blood drawn and fill out a special request form.

## Appendix C. Study 1 Vignettes (Experimental Conditions 1-5)

Duchenne muscular dystrophy (Duchenne's) is a disease that causes muscles to deteriorate and weaken. Duchenne's is a genetic disease and primarily affects boys. The symptoms of Duchenne's develop in early childhood (about age 2-5). Most people with Duchenne's are unable to walk by the time they are a young adult. By the end of their life, most people need respirators to breathe. There is no cure for Duchenne's and most people with Duchenne's die in their 20s or 30s.

Imagine that you or your partner has just given birth to a baby boy. A nurse comes into your room to tell you that there is a new test that screens newborn babies for Duchenne's. Duchenne's cannot be cured and the symptoms cannot be prevented, but the test may help you plan for the future. The test requires a blood sample but causes no long-term harm or side effects. The test will not cost any money.

**[Condition 1 text]** All newborn babies in Michigan are mandated to be screened for certain diseases. In the hospital after birth your baby will have a little blood drawn as part of that screening. Using this blood sample, doctors will test for about 49 of these genetic diseases. The sample can also be used to test for Duchenne's. Adding the screening test for Duchenne's will not require you to come to the hospital at an additional time or draw more blood. The results from the Duchenne's test will be released to you at the same time as the results from the other screening tests.

**[Condition 2 text]** All newborn babies are mandated to be screened for certain diseases. In the hospital after birth your baby will have a little blood drawn as part of that screening. Using this blood sample, doctors will test for about 49 of these genetic diseases. The sample can also be used to test for Duchenne's. Adding the screening test for Duchenne's will not require you to come to the hospital at an additional time or draw more blood. You will receive the results from the Duchenne's test separately from the other results after a short delay.

**[Condition 3 text]** All newborn babies are mandated to be screened for certain diseases. In the hospital after birth your baby will have a little blood drawn as part of that screening. Using this blood sample, doctors will test the following genetic diseases:

**Amino Acid Disorders**
1. Argininemia
2. Argininosuccinic acidemia
3. Citrullinemia
4. Citrullinemia type II
5. Homocystinuria
6. Hypermethioninemia
7. Maple syrup urine disease (MSUD)
8. Phenylketonuria (PKU)
9. Benign hyperphenylalaninemia defect
10. Biopterin cofactor biosynthesis defect
11. Biopterin cofactor regeneration defect
12. Tyrosinemia type I

**Organic Acid Disorders**
26. 2-Methyl-3-hydroxy butyric aciduria
27. 2- Methylbutyryrl-CoA dehydrogenase deficiency
28. 3-Hydroxy 3-methylglutaric aciduria
29. 3-Methylcrotonyl-CoA carboxylase deficiency
30. 3-Methylglutaconic aciduria
31. Beta-ketothiolase deficiency
32. Glutaric acidemia type I
33. Isobutyryl-CoA dehydrogenase deficiency
34. Isovaleric acidemia
35. Malonic acidemia
36. Methylmalonic academia (Cbl A,B)
37. Methylmalonic academia (Cbl C,D)

**Fatty Acid Oxidation Disorders**
13. Carnitine acylcarnitine translocase deficiency
14. Carnitine palmitoyl transferase I deficiency
15. Carnitine palmitoyl transferase II deficiency
16. Carnitine uptake defect
17. Dienoyl-CoA reductase deficiency
18. Glutaric acidemia type II
19. Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency
20. Medium/short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency
21. Medium-chain acyl-CoA dehydrogenase deficiency
22. Medium-chain ketoacyl-CoAthiolase deficiency
23. Short-chain acyl-CoA dehydrogenase deficiency
24. Trifunctional protein deficiency
25. Very long-chain acyl-CoA dehydrogenase deficiency

38. Methylmalonic acidemia (Mutase deficiency)
39. Multiple carboxylase deficiency
40. Propionic acidemia

**Endocrine Disorder**
41. Congenital adrenal
42. Congenital hypothyroidism (CH) hyperplasia (CAH)

**Hemoglobinopathies**
43. S/Beta thalassemia
44. S/C disease
45. Sickle cell anemia
46. Variant hemoglobinopathies

**Other Disorders**
47. Biotinidase deficiency
48. Cystic Fibrosis
49. Galactosemia

The sample can also be used to test for Duchenne's. Adding the screening test for Duchenne's will not require you to come to the hospital at an additional time or draw more blood. The results from the Duchenne's test will be automatically released to you at the same time as the results from the other screening tests.

**[Condition 4 text]** All newborn babies in Michigan are mandated to be screened for certain diseases. In the hospital after birth your baby will have a little blood drawn as part of that screening. Using this blood sample, doctors will test the following genetic diseases:

**Amino Acid Disorders**
1. Argininemia
2. Argininosuccinic acidemia
3. Citrullinemia
4. Citrullinemia type II
5. Homocystinuria
6. Hypermethioninemia
7. Maple syrup urine disease (MSUD)
8. Phenylketonuria (PKU)
9. Benign hyperphenylalaninemia defect
10. Biopterin cofactor biosynthesis defect
11. Biopterin cofactor regeneration defect
12. Tyrosinemia type I

**Fatty Acid Oxidation Disorders**
13. Carnitine acylcarnitine translocase deficiency
14. Carnitine palmitoyl transferase I deficiency
15. Carnitine palmitoyl transferase II deficiency
16. Carnitine uptake defect
17. Dienoyl-CoA reductase deficiency
18. Glutaric acidemia type II
19. Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency
20. Medium/short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency
21. Medium-chain acyl-CoA dehydrogenase deficiency
22. Medium-chain ketoacyl-CoAthiolase deficiency
23. Short-chain acyl-CoA dehydrogenase deficiency
24. Trifunctional protein deficiency
25. Very long-chain acyl-CoA dehydrogenase deficiency

**Organic Acid Disorders**
26. 2-Methyl-3-hydroxy butyric aciduria
27. 2- Methylbutyryrl-CoA dehydrogenase deficiency
28. 3-Hydroxy 3-methylglutaric aciduria
29. 3-Methylcrotonyl-CoA carboxylase deficiency
30. 3-Methylglutaconic aciduria
31. Beta-ketothiolase deficiency
32. Glutaric acidemia type I
33. Isobutyryl-CoA dehydrogenase deficiency
34. Isovaleric acidemia
35. Malonic acidemia
36. Methylmalonic academia (Cbl A,B)
37. Methylmalonic academia (Cbl C,D)
38. Methylmalonic acidemia (Mutase deficiency)
39. Multiple carboxylase deficiency
40. Propionic acidemia

**Endocrine Disorder**
41. Congenital adrenal
42. Congenital hypothyroidism (CH) hyperplasia (CAH)

**Hemoglobinopathies**
43. S/Beta thalassemia
44. S/C disease
45. Sickle cell anemia
46. Variant hemoglobinopathies

**Other Disorders**
47. Biotinidase deficiency
48. Cystic Fibrosis
49. Galactosemia

The sample can also be used to test for Duchenne's. Adding the screening test for Duchenne's will not require you to come to the hospital at an additional time or draw more blood. You will receive the results from the Duchenne's test separately from the other test results after a short delay.

**[Condition 5 text]** The test for Duchenne's can be done in the hospital after birth. The results from the test will be automatically released to you.

## Appendix D. Study 1 Brochures

Brochure: Bundled Mandatory NBS Panel

**Do I Want My Newborn Son Screened for Duchenne Muscular Dystrophy?**

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear until age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**Why might I want my son screened for DMD?**

- Without screening it might take years before DMD is diagnosed.
- Services like physical therapy can be started early.
- Treatment can slow down the progress of DMD.
- Knowing if your son has DMD can help you plan for the future.

**Why might I *not* want my son screened for DMD?**

- Although treatments are available, there is *no* cure.
- An early diagnosis could change how you treat your son.
- You might want to think your son is healthy until symptoms begin.

**How is DMD screening done?**

- *All* babies have a few drops of blood taken from their heel after birth.
- This blood is used to test for 50 genetic diseases that can be treated early to prevent serious problems or death.
- This test is called Newborn Screening (NBS).
- The *same* blood sample can be used to screen for DMD. You will not have to come to the hospital at an additional time or draw more blood.

Brochure: Unbundled Mandatory NBS Panel

## Do I Want My Newborn Son Screened for Duchenne Muscular Dystrophy?

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear until age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**Why might I want my son screened for DMD?**

- Without screening it might take years before DMD is diagnosed.
- Services like physical therapy can be started early.
- Treatment can slow down the progress of DMD.
- Knowing if your son has DMD can help you plan for the future.

**Why might I *not* want my son screened for DMD?**

- Although treatments are available, there is *no* cure.
- An early diagnosis could change how you treat your son.
- You might want to think your son is healthy until symptoms begin.

**How is DMD screening done?**

- *All* babies have a few drops of blood taken from their heel after birth.
- This blood is used to test for 50 genetic diseases that can be treated early to prevent serious problems or death.
- This test is called Newborn Screening (NBS).
- The *same* blood sample can be used to screen for DMD. You will not have to come to the hospital at an additional time or draw more blood.

### Newborn Screening Tests

**Amino Acid Disorders**
1. Argininemia
2. Argininosuccinic acidemia
3. Citrullinemia Type I, II
4. Homocystinuria
5. Hypermethioninemia
6. Maple syrup urine disease (MSUD)
7. Phenylketonuria (PKU)
8. Benign hyperphenylalaninemia defect
9. Biopterin cofactor biosynthesis defect
10. Biopterin cofactor regeneration defect
11. Tyrosinemia Type I, II, III

**Fatty Acid Oxidation Disorders**
12. Carnitine acylcarnitine translocase deficiency
13. Carnitine palmitoyl transferase I deficiency
14. Carnitine palmitoyl transferase II deficiency
15. Carnitine uptake defect
16. Dienoyl-CoA reductase deficiency
17. Glutaric acidemia type II
18. Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency
19. Medium/short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency
20. Medium-chain acyl-CoA dehydrogenase deficiency
21. Medium-chain ketoacyl-CoAthiolase deficiency
22. Short-chain acyl-CoA dehydrogenase deficiency
23. Trifunctional protein deficiency
24. Very long-chain acyl-CoA dehydrogenase deficiency

**Hemoglobinopathies**
25. S/Beta thalassemia
26. S/C disease
27. Sickle cell anemia
28. Variant hemoglobinopathies
29. Hemoglobin H disease

**Organic Acid Disorders**
30. 2-Methyl-3-hydroxy butyric aciduria
31. 2- Methylbutyryrl-CoA dehydrogenase deficiency
32. 3-Hydroxy 3-methylglutaric aciduria
33. 3-Methylcrotonyl-CoA carboxylase deficiency
34. 3-Methylglutaconic aciduria
35. Beta-ketothiolase deficiency
36. Glutaric acidemia type I
37. Isobutyryl-CoA dehydrogenase deficiency
38. Isovaleric acidemia
39. Methylmalonic acidemia cobalamin disorders (Cbl A,B)
40. Methylmalonic aciduria with homocystinuria (Cbl C,D)
41. Methylmalonic acidemia methylmalonyl-CoA mutase
42. Multiple carboxylase deficiency
43. Propionic acidemia

**Endocrine Disorder**
44. Congenital adrenal hyperplasia (CAH)
45. Congenital hypothyroidism (CH)

**Other Disorders**
46. Biotinidase deficiency
47. Galactosemia (GAL)
48. Cystic Fibrosis (CF)
49. T-cell related lymphocyte deficiencies
50. Severe combined immunodeficiency (SCID)

**Optional Tests**
Duchenne muscular dystrophy (DMD)

Brochure: No Mandatory NBS Panel

## Do I Want My Newborn Son Screened for Duchenne Muscular Dystrophy?

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear until age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**Why might I want my son screened for DMD?**

- Without screening it might take years before DMD is diagnosed.
- Services like physical therapy can be started early.
- Treatment can slow down the progress of DMD.
- Knowing if your son has DMD can help you plan for the future.

**Why might I *not* want my son screened for DMD?**

- Although treatments are available, there is *no* cure.
- An early diagnosis could change how you treat your son.
- You might want to think your son is healthy until symptoms begin.

**How is DMD screening done?**

- DMD screening will not require you to come to the hospital at an additional time.

**Appendix E. Survey Measures**

| Measure | Item/Scale | Response Options | Citation |
|---|---|---|---|
| *Primary outcome* | | | |
| DMD test utilization | How likely do you think that it is that you will choose to have your baby tested for DMD? | 0=Very unlikely<br>1=Unlikely<br>2=Likely<br>3=Very likely | Created |
| *Attitudes & beliefs* | | | |
| DMD NBS - importance | How important is it that your baby is tested for DMD? | 0=Not at all important<br>1<br>2<br>3<br>4=Very important | Created |
| DMD NBS - importance | How important is it that you see the results from your baby's test for DMD? | 0=Not at all important<br>1<br>2<br>3<br>4=Very important | Created |
| DMD NBS - importance | How important is it to share the results from the DMD test with others? | 0=Not at all important<br>1<br>2<br>3<br>4=Very important | Created |
| DMD NBS - impact | How much do you think you would worry about the results of your baby's DMD test? | 0=Not at all<br>1<br>2<br>3<br>4=Very Much | Created |

| | | | |
|---|---|---|---|
| DMD NBS - impact | The information from the DMD test may help me prepare for the future. | 0=Strongly disagree<br>1<br>2<br>3<br>4=Strongly agree | Created |
| DMD NBS - impact | The information from the DMD test would affect whether I have more children. | 0=Strongly disagree<br>1<br>2<br>3<br>4=Strongly agree | Created |
| DMD NBS - impact | The results from the DMD test may affect how I treat my child. | 0=Strongly disagree<br>1<br>2<br>3<br>4=Strongly agree | Created |
| DMD NBS - impact | My child would be treated differently by others if he is diagnosed with DMD. | 0=Strongly disagree<br>1<br>2<br>3<br>4=Strongly agree | Created |
| NBS in general | Do you think your child will be treated differently by others if he has an incurable disease? | 0=Strongly disagree<br>1<br>2<br>3<br>4=Strongly agree | Created |
| NBS in general | Do you think having a child with an incurable disease changes how you might treat your child? | 0=Strongly disagree<br>1<br>2<br>3<br>4=Strongly agree | Created |
| Information | How important is it that you have all the information available about your child? | 0=Not at all important<br>1 | Created |

| | | | |
|---|---|---|---|
| | | 2<br>3<br>4=Very important | |
| Subjective norm re: newborn screening (direct measure) | Do you think that most people agree or disagree that it is important for babies to be tested for as many genetic diseases as possible? | -3 = Disagree<br>-2<br>-1<br>0<br>1<br>2<br>3=agree | Theory of Reasoned Action |
| Subjective norm re: DMD test (direct measure) | Do you think that most people agree or disagree with getting the DMD test? | -3 = Disagree<br>-2<br>-1<br>0<br>1<br>2<br>3=agree | Theory of Reasoned Action |
| *Subject characteristics* | | | |
| Age | What is your age? | Open ended | |
| Gender | What is your gender? | Male; Female | |
| Educational attainment | What is the highest level of education that you have received? | Less than high school; High school graduate; College graduate; More than college | |
| Marital status | What is your marital status? | Married/partnered;<br>Not married/partnered | |
| Household income | Including all sources of income, what is your total household income? | Less than $14,500;<br>$14,500- less than $35,000;<br>$35,000- less than $50,000;<br>$50,000-less than $75,000;<br>$75,000-less than $100,000;<br>$100,000 and over | National Health Interview Survey (Centers for Disease Control and Prevention) |
| Race | What is your race? (select all that apply) | White; African American/Black; Native | National Health Interview Survey |

| | | | |
|---|---|---|---|
| | | Hawaiian and Other Pacific Islander; Asian; American Indian/Alaska Native | (Centers for Disease Control and Prevention) |
| Experience in the health field | What is your experience working in the health or research field? | Currently working in the health or research field; Previously worked in the health or research field; Never worked in the health or research field | Created |
| Pregnancy history | Have you or your partner ever been pregnant? | Yes; No; Don't know | Created |
| Pregnancy history | How many children have you or your partner had? | Open ended | |
| History of pregnancies with complications | Have you had any complications during a pregnancy? | Yes; No; Don't know | Created |
| Prenatal screening history | Have you ever had any prenatal screening tests done during a pregnancy? | Yes; No; Don't know | Created |
| Newborn screening history | Did your child/children have newborn screening tests done? | Yes; No; Don't know | Created |
| Child health history | Has your child/children had any illnesses? | Acute illness; Chronic illness; Both an acute illness and a chronic illness; No; Don't know | Created |
| Experience with people with chronic illnesses | Do you know someone with a chronic illness? | Yes; No; Don't know | Created |
| Familiarity with the disease | Do you know of a family member or friend diagnosed with DMD or another genetic disorder? | Yes, a family member; Yes, a friend; No; Don't know | Created |
| *Individual differences* | | | |
| Cognitive Style | Please answer the following questions about yourself:<br>1. I like to have the responsibility of handling a situation that requires a lot of thinking.<br>2. Thinking is not my idea of fun. (*reverse coded*) | 1=Strongly Disagree<br>2=Disagree<br>3=Neither Agree nor Disagree<br>4=Agree | *Need for Cognition* Cacioppo et al. (1984) |

| | | | |
|---|---|---|---|
| | 3. I would rather do something that requires little thought than something that is sure to challenge my thinking abilities. (*reverse coded*)<br>4. The idea of relying on thought to make my way to the top appeals to me.<br>5. I really enjoy a task that involves coming up with new solutions to problems.<br>6. Learning new ways to think doesn't excite me very much. (*reverse coded*)<br>7. The notion of thinking abstractly is appealing to me. | 5=Strongly Agree<br><br>The final score is a sum of each of the 7 items | |
| Monitoring/blunting coping style | Different people tend to respond in different ways when faced with difficult or threatening situations. The following question describes a possible difficult situation which you may encounter. Please consider each scenario and indicate how you think you would react.<br><br>Vividly imagine that you are afraid of the dentist and have to get some dental work done. Which of the following would you do? Tick all of the statements that might apply to you:<br>1. I would ask the dentist exactly what he was going to do. (M)<br>2. I would take a tranquilizer or have a drink before going. (B)<br>3. I would try to think about pleasant memories. (B)<br>4. I would want the dentist to tell me when I would feel pain. (M)<br>5. I would try to sleep. (B) | Items are marked as Monitoring (M) or Blunting (B). To obtain the total score, add up all the M scores and B scores and subtract the Total B score from the Total M. The higher (more positive) the score, the greater the monitoring. | *Miller Behavioral Style Scale*<br>Miller (1987) |

| | | | |
|---|---|---|---|
| | 6. I would watch all the dentist's movements and listen for the sound of the drill. (M)<br>7. I would watch the flow of water from my mouth to see if it contained blood. (M)<br>8. I would do mental puzzles in my mind. (B) | | |
| Altruism | Check the category on the right that conforms to the frequency with which you have carried out the following acts:<br>1. I have donated goods or clothes to a charity.<br>2. I have done volunteer work for a charity.<br>3. I have delayed an elevator and held the door open for a stranger.<br>4. I have allowed someone to go ahead of me in a line (e.g., supermarket, copying machine, etc.).<br>5. I have bought "charity" Christmas cards deliberately because I knew it was for a good cause.<br>6. I have helped a classmate who I did not know that well with a homework assignment when my knowledge was greater than his or hers.<br>7. I have, before being asked, voluntarily looked after a neighbor's pets or children without being paid for it.<br>8. I have offered to help a handicapped or elderly stranger across a street. | 0=Never<br>1<br>2<br>3<br>4=Very often<br><br>An overall score is calculated by averaging the items. | *Altruism Scale* (Safe Altruism subscale) Homant (2010) |
| Privacy | Here are some statements about personal information. From the standpoint of personal privacy, please indicate the extent to which you, as an individual, agree or disagree with each statement.<br>1. It usually bothers me when companies ask me for personal information. | 0=Strongly Disagree<br>1<br>2<br>3<br>4<br>5 | *Concern for Information Privacy Instrument* Smith et al (1996) |

| | | | |
|---|---|---|---|
| | 2. When companies ask me for personal information, I sometimes think twice before providing it.<br>3. It bothers me to give personal information to so many companies.<br>4. I am concerned that companies are collecting too much personal information about me. | 6=Strongly Agree<br><br>An overall score is calculated by averaging the items. | |
| Medical Distrust | The next questions are about your opinion of the health care system in general. When we refer to the health care system, we mean hospitals, health insurance groups, and medical research. For each statement below, please check how strongly you agree or disagree:<br>1. Medical experiments can be done on me without my knowing about it.<br>2. My medical records are kept private. (*reverse coded*)<br>3. People die every day because of mistakes by the health care system.<br>4. When they take my blood, they do tests they don't tell me about.<br>5. If a mistake were made in my health care, the health care system would try to hide it from me.<br>6. People can get access to my medical records without my approval.<br>7. The health care system cares more about holding costs down than it does about doing what is needed for my health.<br>8. I receive high-quality medical care from the health care system. (*reverse coded*)<br>9. The health care system puts my medical needs | 0=Strongly Disagree<br>1=Disagree<br>2=Not Sure<br>3=Agree<br>4=Strongly Agree<br><br>An overall score is calculated by averaging the items. | *Health Care System Distrust Scale*<br>Rose et al (2004) |

| | | | |
|---|---|---|---|
| | above all other considerations when treating my medical problems. (*reverse coded*)<br>10. Some medicines have things in them that they don't tell you about. | | |
| Government Distrust | How much do you agree or disagree with the following statements?<br>1.When there is a really serious health or environmental problem, then public officials will take care of it.<br>2.Until they alert me about a specific problem, I don't really have to worry.<br>3.I have very little control over risks to my health.<br>4.Those in power often withhold information about things that are harmful to us.<br>5.The land, air, and water around us are, in general, more contaminated now than ever before.<br>6.Continued economic growth can only lead to pollution and depletion of natural resources. | 0=Strongly Agree<br>1=Agree<br>2=Disagree<br>3=Strongly Disagree<br><br>An overall score is calculated by averaging the items. | Peters & Slovic (1996) |

## Appendix F. Study 2 Vignettes (Experimental Conditions 1-5)

Duchenne muscular dystrophy (Duchenne's) is a disease that causes muscles to deteriorate and weaken. Duchenne's is a genetic disease and primarily affects boys. The symptoms of Duchenne's develop in early childhood (about age 2-5). Most people with Duchenne's are unable to walk by the time they are a young adult. By the end of their life, most people need respirators to breathe. There is no cure for Duchenne's and most people with Duchenne's die in their 20s or 30s.

Imagine that you or your partner has just given birth to a baby boy. A nurse comes into your room to tell you that there is a new test that screens newborn babies for Duchenne's. Duchenne's cannot be cured and the symptoms cannot be prevented, but the test may help you plan for the future. The test requires a blood sample but causes no long-term harm or side effects. The test will not cost any money.

**[Condition 1 text]** If you choose to have the test done, the hospital nurse will draw a blood sample which will then be sent to a private testing company to be tested for Duchenne's. The test results will be given to you personally, which means that the results will not be entered into your baby's medical record and will not be available to doctors, or anyone else, unless you grant them access.

**[Condition 2 text]** If you choose to have the test done, the hospital nurse will draw a blood sample, which will then be tested for Duchenne's. The test results will be entered into your baby's confidential personal health record, which means that only people with access to your baby's personal health record will be able to see the results.

**[Condition 3 text]** If you choose to have the test done, the hospital nurse will draw a blood sample, which will then be tested for Duchenne's. The test results will be entered into your baby's personal health record, which means that only people with access to your baby's personal health record will be able to see the results. You can also choose to have part of the blood sample be stored in the hospital to be used in future medical research studies which may develop new testing and treatment options for Duchenne's.

**[Condition 4 text]** If you choose to have the test done, the hospital nurse will draw a blood sample, which will then be tested for Duchenne's. The test results will be entered into your baby's confidential personal health record, which means that only people with access to your baby's personal health record will be able to see the results. You can also choose to have the results entered into a national registry specifically of newborns with Duchenne's, which may lead to a better understanding of Duchenne's testing and treatment options for children living with Duchenne's, including possibly your child.

**[Condition 5 text]** If you choose to have the test done, the hospital nurse will draw a blood sample, which will then be tested for Duchenne's. The test results will be entered into your baby's confidential personal health record, which means that only people with access to your baby's personal health record will be able to see the results. You can also choose to have the results entered into a national registry of all newborns, which follow trends in children's health and lead to a better understanding of general medical care.

## Appendix G. Study 2 Brochures

Brochure: Private Release

### Do I Want My Newborn Son Screened for Duchenne Muscular Dystrophy?

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear until age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**Why might I want my son screened for DMD?**

- Without screening it might take years before DMD is diagnosed.
- Services like physical therapy can be started early.
- Treatment can slow down the progress of DMD.
- Knowing if your son has DMD can help you plan for the future.

**Why might I *not* want my son screened for DMD?**

- Although treatments are available, there is *no* cure.
- An early diagnosis could change how you treat your son.
- You might want to think your son is healthy until symptoms begin.

**How is DMD screening done?**

- *All* babies have a few drops of blood taken from their heel after birth.
- This blood is used to test for genetic diseases that can be treated early to prevent serious problems or death.
- This test is called Newborn Screening (NBS).
- The *same* blood sample can be used to screen for DMD.

**Who will learn the results of the DMD test?**

- The results will *only* be given to you and will not be shared with *anyone else*, unless you give them permission.

**How much does DMD screening cost?**

- DMD screening is *free*.

Brochure: Electronic Health Record Release

## Do I Want My Newborn Son Screened for Duchenne Muscular Dystrophy?

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear until age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**Why might I want my son screened for DMD?**

- Without screening it might take years before DMD is diagnosed.
- Services like physical therapy can be started early.
- Treatment can slow down the progress of DMD.
- Knowing if your son has DMD can help you plan for the future.

**Why might I *not* want my son screened for DMD?**

- Although treatments are available, there is *no* cure.
- An early diagnosis could change how you treat your son.
- You might want to think your son is healthy until symptoms begin.

**How is DMD screening done?**

- *All* babies have a few drops of blood taken from their heel after birth.
- This blood is used to test for genetic diseases that can be treated early to prevent serious problems or death.
- This test is called Newborn Screening (NBS).
- The *same* blood sample can be used to screen for DMD.

**Who will learn the results of the DMD test?**

- The results will be entered into your baby's health record.
- Only you and your child's current or future health providers will learn the results.

**How much does DMD screening cost?**

- DMD screening is *free*.

Brochure: Biobank Release

## Do I Want My Newborn Son Screened for Duchenne Muscular Dystrophy?

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear until age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**Why might I want my son screened for DMD?**

- Without screening it might take years before DMD is diagnosed.
- Services like physical therapy can be started early.
- Treatment can slow down the progress of DMD.
- Knowing if your son has DMD can help you plan for the future.

**Why might I *not* want my son screened for DMD?**

- Although treatments are available, there is *no* cure.
- An early diagnosis could change how you treat your son.
- You might want to think your son is healthy until symptoms begin.

**How is DMD screening done?**

- *All* babies have a few drops of blood taken from their heel after birth.
- This blood is used to test for genetic diseases that can be treated early to prevent serious problems or death.
- This test is called Newborn Screening (NBS).
- The *same* blood sample can be used to screen for DMD.

**Who will learn the results of the DMD test?**

- The results will be entered into your baby's health record, so only you and your child's current or future health providers will learn the results.
- Medical researchers at this or other hospitals may be able to learn the results as part of approved research projects.
- The goal of this research is to develop new testing and treatment options for DMD.

**How much does DMD screening cost?**

- DMD screening is *free*.

Brochure: DMD Registry Release

## Do I Want My Newborn Son Screened for Duchenne Muscular Dystrophy?

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear until age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**Why might I want my son screened for DMD?**

- Without screening it might take years before DMD is diagnosed.
- Services like physical therapy can be started early.
- Treatment can slow down the progress of DMD.
- Knowing if your son has DMD can help you plan for the future.

**Why might I *not* want my son screened for DMD?**

- Although treatments are available, there is *no* cure.
- An early diagnosis could change how you treat your son.
- You might want to think your son is healthy until symptoms begin.

**How is DMD screening done?**

- *All* babies have a few drops of blood taken from their heel after birth.
- This blood is used to test for genetic diseases that can be treated early to prevent serious problems or death.
- This test is called Newborn Screening (NBS).
- The *same* blood sample can be used to screen for DMD.

**Who will learn the results of the DMD test?**

- People with access to your baby's health record, like current or future health providers, will learn the results.
- The results will also be entered into a registry that collects medical data to track the health of *children with DMD*.
- This DMD registry may lead to new testing and treatment options for children with DMD, including possibly your child.

**How much does DMD screening cost?**

- DMD screening is *free*.

Brochure: General Registry Release

## Do I Want My Newborn Son Screened for Duchenne Muscular Dystrophy?

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear until age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**Why might I want my son screened for DMD?**

- Without screening it might take years before DMD is diagnosed.
- Services like physical therapy can be started early.
- Treatment can slow down the progress of DMD.
- Knowing if your son has DMD can help you plan for the future.

**Why might I *not* want my son screened for DMD?**

- Although treatments are available, there is *no* cure.
- An early diagnosis could change how you treat your son.
- You might want to think your son is healthy until symptoms begin.

**How is DMD screening done?**

- *All* babies have a few drops of blood taken from their heel after birth.
- This blood is used to test for genetic diseases that can be treated early to prevent serious problems or death.
- This test is called Newborn Screening (NBS).
- The *same* blood sample can be used to screen for DMD.

**Who will learn the results of the DMD test?**

- People with access to your baby's health record, like current or future health providers, will learn the results.
- The results will also be entered into a registry that collects medical data to track trends in the health of *all children*.
- This registry may lead to a new understanding of overall medical care and healthcare services.

**How much does DMD screening cost?**

- DMD screening is *free*.

## Appendix H. Study 3 Vignettes (Experimental Conditions 1-8)

Duchenne muscular dystrophy (Duchenne's) is a disease that causes muscles to deteriorate and weaken. Duchenne's is a genetic disease and primarily affects boys. The symptoms of Duchenne's develop in early childhood (about age 2-5). Most people with Duchenne's are unable to walk by the time they are a young adult. By the end of their life, most people need respirators to breathe. There is no cure for Duchenne's and most people with Duchenne's die in their 20s or 30s.

**[Condition 1 text]** Imagine you or your partner gave birth to a baby boy yesterday. He was born full-term at 40 weeks and is a healthy 20 inches and 8 pounds. Your doctor has no concerns about his immediate health. You and your son will be released from the hospital tomorrow.

While you are sitting in your hospital room, a nurse comes into your room to discuss a type of test for newborn babies that the hospital is offering are offering parents. She tells you that a new test has been developed that screens newborn babies for Duchenne's Muscular Dystrophy (DMD) and gives you information about the test.

**[Condition 2 text]** Imagine you or your partner gave birth to a baby boy yesterday. He was born full-term at 40 weeks and is a healthy 20 inches and 8 pounds. Your doctor has no concerns about his immediate health. You and your son will be released from the hospital tomorrow.

One thing you are concerned about is your family history of a genetic disorder called Duchenne's Muscular Dystrophy (DMD). Your uncle (on your mother's side) was diagnosed with DMD at age 3 and by age 12 was wheelchair dependent due to muscle deterioration. Your uncle was eventually paralyzed and died from DMD at age 20.

While you are sitting in your hospital room, a nurse comes into your room to discuss a type of test for newborn babies that the hospital is offering are offering parents. She tells you that a new test has been developed that screens newborn babies for DMD and gives you information about the test.

**[Condition 3 text]** Imagine you or your partner gave birth to a baby boy yesterday. He was born full-term at 40 weeks and is a healthy 20 inches and 8 pounds. Your doctor has no concerns about his immediate health. You and your son will be released from the hospital tomorrow.

One thing you are concerned about is your family history of Epilepsy, a neurological disorder that causes seizures. Both your father and your father's sister have a medical history of having seizures.

While you are sitting in your hospital room, a nurse comes into your room to discuss a type of test for newborn babies that the hospital is offering are offering parents. She tells you that a new test has been developed that screens newborn babies for Duchenne's Muscular Dystrophy (DMD) and gives you information about the test.

**[Condition 4 text]** Imagine you or your partner gave birth to a baby boy yesterday. He was born prematurely, at 33 weeks instead of the expected 40. He is 17 inches and 4 pounds, compared to full-term babies' average size of 20 inches and 8 pounds. . Because your son was born prematurely, your doctor has important concerns about his immediate health. You will be able to leave the hospital tomorrow, but your son will need to stay in the hospital for another week.

There are several issues with your son that you are concerned about because of his premature birth. In particular, he has difficulty feeding and sometimes needs to be fed through a temporary tube through his mouth. You know that he will need to be able to feed without help before he can go home. He will also have to be followed for a few years to make sure that he does not develop any cognitive or physical developmental problems.

While you are sitting in your hospital room, a nurse comes into your room to discuss a type of test for newborn babies that the hospital is offering are offering parents. She tells you that a new test has been developed that screens newborn babies for Duchenne's Muscular Dystrophy (DMD), a genetic condition that causes all muscles in the body to become weak over time, and gives you information about the test.

**[Condition 5 text]** Imagine you or your partner gave birth to a baby boy yesterday. He was born full-term at 40 weeks and is a healthy 20 inches and 8 pounds. Your doctor has no concerns about his immediate health. You and your son will be released from the hospital tomorrow.

While you are sitting in your hospital room, a nurse comes into your room to discuss that researchers at a local university are offering parents. She tells you that a new test has been developed that screens newborn babies for Duchenne's Muscular Dystrophy (DMD) and gives you information about the test.

**[Condition 6 text]** Imagine you or your partner gave birth to a baby boy yesterday. He was born full-term at 40 weeks and is a healthy 20 inches and 8 pounds. Your doctor has no concerns about his immediate health. You and your son will be released from the hospital tomorrow.

One thing you are concerned about is your family history of a genetic disorder called Duchenne's Muscular Dystrophy (DMD). Your uncle (on your mother's side) was diagnosed with DMD at age 3 and by age 12 was wheelchair dependent due to muscle deterioration. Your uncle was eventually paralyzed and died from DMD at age 20.

While you are sitting in your hospital room, a nurse comes into your room to discuss that researchers at a local university are offering parents. She tells you that a new test has been developed that screens newborn babies for DMD and gives you information about the test.

**[Condition 7 text]** Imagine you or your partner gave birth to a baby boy yesterday. He was born full-term at 40 weeks and is a healthy 20 inches and 8 pounds. Your doctor has no concerns about his immediate health. You and your son will be released from the hospital tomorrow.

One thing you are concerned about is your family history of Epilepsy, a neurological disorder that causes seizures. Both your father and your father's sister have a medical history of having seizures.

While you are sitting in your hospital room, a nurse comes into your room to discuss that researchers at a local university are offering parents. She tells you that a new test has been developed that screens newborn babies for Duchenne's Muscular Dystrophy (DMD) and gives you information about the test.

**[Condition 8 text]** Imagine you or your partner gave birth to a baby boy yesterday. He was born prematurely, at 33 weeks instead of the expected 40. He is 17 inches and 4 pounds, compared to full-term babies' average size of 20 inches and 8 pounds. . Because your son was born prematurely, your doctor has important concerns about his immediate health. You will be able to leave the hospital tomorrow, but your son will need to stay in the hospital for another week.

There are several issues with your son that you are concerned about because of his premature birth. In particular, he has difficulty feeding and sometimes needs to be fed through a temporary tube through his mouth. You know that he will need to be able to feed without help before he can go home. He will also have to be followed for a few years to make sure that he does not develop any cognitive or physical developmental problems.

While you are sitting in your hospital room, a nurse comes into your room to discuss that researchers at a local university are offering parents. She tells you that a new test has been developed that screens newborn babies for Duchenne's Muscular Dystrophy (DMD), a genetic condition that causes all muscles in the body to become weak over time, and gives you information about the test.

## Appendix I. Study 3 Brochures

Brochure: Personal Purpose for DMD NBS

**Do I Want My Newborn Son Screened for Duchenne Muscular Dystrophy?**

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**What is newborn screening?**

- *All* babies have a few drops of blood taken from their heel after birth.
- This blood is used to test for other genetic diseases that *can* be treated early to prevent serious problems or death.
- This test is called Newborn Screening (NBS).

**What is the test for DMD?**

- An additional test was developed for DMD in newborns.
- This test uses the same blood sample as standard NBS.

**Why might I want my son screened for DMD?**

- Without screening it might take years before DMD is diagnosed.
- Treatments like physical therapy can be started early and delay DMD progress.
- Knowing if your son has DMD can help you plan for the future.
- The test results will also be entered into the hospital database for future approved research projects to develop new testing and treatment options for DMD.
- You may want to help with these research goals to help other children.

**Why might I *not* want my son screened for DMD?**

- Although some treatments are available, there is *no* cure.
- An early diagnosis of DMD could change how you treat your son.
- You might want to think your son is healthy until symptoms begin.
- You may not want your son's information to be added to a research database and used by people other than your doctors.

**How much does DMD screening cost?**

- Nothing. DMD screening is *free*.

Brochure: Research Purpose for DMD NBS

**Do I Want My Newborn Son To Participate in Research That Will Screen Him for Duchenne Muscular Dystrophy?**

**What is Duchenne (Doo-shen) muscular dystrophy (DMD)?**

- DMD is a genetic condition that causes all muscles in the body to become weak over time.
- Clear signs of DMD are not present at birth, and usually don't appear until age 2-3.
- Boys with DMD might walk later than other boys and eventually boys with DMD need a wheelchair.
- Boys with DMD have a shortened lifespan.
- Treatment can slow down the disease, but there is *no* cure.

**What is this research?**

- Public health researchers are trying to identify newborns with DMD.
- Parents who participate in this study will have their sons tested for DMD.
- The goal of this research is to develop new testing and treatment options for DMD.
- If you are willing to participate, as a part of this research you will also be given the results of the test.

**What will my son do to participate in this research?**

- *All* babies have a few drops of blood taken from their heel after birth.
- This blood is used to test for other genetic diseases that *can* be treated early to prevent serious problems or death.
- This test is called Newborn Screening (NBS).
- The testing for DMD to be done in this research study uses the same blood sample as is used for the required newborn screening.
- The test results are entered into a database used for approved research projects associated with DMD. They are not entered into your medical record.

**Why might I agree to participate in this research?**

- Contributing to this research will help to identify children with DMD and learn which types of children are most at risk for DMD.
- You will learn whether your son has DMD. Without screening it might take years before DMD is diagnosed.
- Treatments like physical therapy can be started early and delay DMD progress.
- Knowing whether your son has DMD can help you plan for the future.

**Why might I *not* agree to participate in this research?**

- Participating in this research is optional. You do not have to participate.
- You may not want your son's information to be added to a research database and used by people other than your doctors.
- An early diagnosis of DMD could change how you treat your son.
- Although treatments are available, there is *no* cure.
- You might want to think your son is healthy until symptoms begin.

**How much does this research study cost?**

- Nothing. Receiving DMD screening through this research project is *free*.

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