Developing a Conjoint Analysis Survey of Parental Attitudes Regarding Voluntary Newborn Screening

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Contents

Abstract

Newborn screening for genetic conditions is conducted in all 50 states, but parents' opinions of such screening are largely unknown. As newborn screening has expanded from a few relatively common conditions requiring early treatment to a broader spectrum of conditions with uncertain prognosis, it is important to understand parents' views. Stated preference surveys provide quantifiable data on parent preferences about features of newborn screening tests, the economic value of testing, and the effect of a test's features on its probable uptake and value.

We conducted formative research and developed a stated preference survey on parents' preferences regarding voluntary newborn screening. We reviewed the literature on parents' attitudes toward newborn screening and factors related to those attitudes and developed a list of condition and test attributes. We narrowed the list by expert review and conducted focus groups with parents of infants to determine if they understood the attributes and to see which attributes parents viewed as relevant.

We found that some parents struggled with the probability-based attributes. The most relevant attributes were developmental disability, physical disability, incidence, recurrence, lifespan, sensitivity, and cost. The survey developed from this study could provide data for economic evaluation, test prioritization, and educational material development.

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Introduction

Conjoint analysis, also known as a "stated choice" or a "discrete choice" experiment, is a form of stated preference research based on the economic theory of choice data. It assumes that a good (e.g., test, product, policy) is composed of various attributes and that an individual's utility or satisfaction is a function of these attributes.[1] Any attribute that varies between decision-making contexts can be included, but attributes that are key to policy making are given priority. Cognitive research suggests that a survey should have fewer than 10 attributes and ideally between 5 and 7 attributes. Less important attributes are included as invariant features discussed in information provided alongside of or before the trade-off exercises, so these attributes are reflected in the estimated uptake levels and the value of the test. Individual preferences regarding these attributes are elicited by presenting participants with a series of choices about two or more hypothetical tests with different attributes (called trade-offs).

We expect that the relative importance of attributes will vary in a sample and that individuals will accept trade-offs between different features. Individuals may be willing to accept a less desirable level of a less important attribute to achieve the preferred level of a more important attribute. Conjoint analysis thus parallels consumers' everyday decision making and is more natural than other stated preference methods, such as contingent valuation, in which respondents are asked how much they would be "willing to pay" for a good.

Conjoint analysis has been used widely in market research, environmental economics, and transportation economics and, more recently, in health economics and public health.[2,3] Although typical quantitative analysis requires data on observed choices (i.e., revealed preference data), stated preference surveys, including conjoint analysis, can be used when observed data do not exist, as with new technologies such as newborn screening. Conjoint analysis also can be used to estimate uptake and acceptability for programs, policies, goods, and tests. Although conjoint analysis has been very influential outside of health care (see Louviere et al.[1] for more discussion), its influence in health care and health policy is still in its relative infancy,

lagging its use in economics overall by a few years. Pharmacoeconomics—economic analysis applied to evaluation of pharmaceuticals and medical technology—has been very accepting of conjoint analysis, both for quantifying benefits and for identifying patient preferences for treatment. Bridges et al.[2] provide an excellent survey of the uses of conjoint analysis in pharmoeconomics.

Public health acceptance of conjoint analysis is high in the United Kingdom and Europe, as indicated by several important and influential studies of health care and health reform,[4] physician choice,[5] and women's health.[6] Indeed, the seminal general audience article on conjoint analysis by Ryan and Farrar[7] was published in a leading UK medical journal, and their conjoint analysis work was originally spurred by government requests for improved data on women's health treatment in Scotland.[6] The presence of government-policy organizations such as the UK National Institute for Health and Clinical Excellence, which requires economic data, has provided an audience of policy makers as well as academics for conjoint analysis studies. As the method expands internationally and in the United States, conjoint analysis may be expected to have greater influence in public health internationally as well.

Newborn genetic screening is well established. Public health programs that screen newborn infants for genetic conditions were established in the 1960s, when three states (New York, Pennsylvania, and Ohio) passed laws mandating screening for phenylketonuria (PKU).[8] Newborn screening for genetic conditions is now routine in all 50 states and is mandatory in most states.[9] Even in the few states that allow parents to opt out of screening, parental acceptance is over 90 percent.[10-12] However, the context of newborn screening has changed in the past decade with scientific and technological advances.

Until recently, newborn screening was limited to a few relatively common conditions that could be treated effectively before symptom onset. The number of conditions was limited by the cost of testing and follow-up and by the small amount of blood available from a capillary heel stick, the usual collection method. Two developments changed the screening process: tandem mass spectrometry

and gene sequencing. Tandem mass spectrometry enabled screening for many additional conditions using the same sample size and with minimal increase in the cost of testing. The recent availability and rapidly decreasing cost of gene-sequencing tests further expand the number of conditions for which testing is available.[13] With these new technologies, testing can be offered for conditions that have no effective treatment or that can be treated successfully after symptoms appear.[13,14] Some professionals and parents promote newborn screening for such conditions if early diagnosis benefits the family. For example, many advocates of expanded newborn screening are parents of children affected with conditions such as fragile X, for which presymptomatic diagnosis is now available.[15] The expected benefits of expanded screening cited by parents and screening advocates include reducing the time and resources spent during the diagnosis process and providing information for reproductive decision making.[16,17]

Little is known about the views of parents of healthy newborns on the value of current screening practices, much less the benefits and harms of expanding newborn screening to include tests with no immediate medical benefit. Most research focuses on the reactions of parents of children affected with a condition or on parents whose children have had a false-positive result.[18] A recent review of the psychosocial aspects of newborn and antenatal screening found only 28 articles that examined newborn screening, compared with 78 on antenatal screening.[18] Perhaps research is not often conducted on parental attitudes toward newborn screening because there is little opposition to the practice, which suggests that most parents and physicians view current newborn screening as unquestionably beneficial. Critics of expanded newborn screening are concerned about the potential negative effects of false-positive tests or of presymptomatic diagnosis and about the violation of a child's autonomy.[14] To address these concerns, some advocates propose voluntary screening.[19] It is not clear, however, how parents will view expanded screening, be it voluntary or mandatory.

Conjoint analysis stated preference surveys can identify and clarify parents' attitudes toward presymptomatic newborn screening by allowing us to quantify their preferences for specific features of newborn screening tests and to quantify the economic value (amount parents would be willing to pay) of testing.[20] This method also provides data that help us estimate parental acceptance (uptake) of the tests and understand how different features of the tests would affect parental acceptance and the economic value of the test. It can also identify the relative importance of test characteristics and quantify them in terms of willingness to pay. Only very limited data are available on the economic value of the benefits of expanded screening, the costs of the negative consequences of screening, or its probable use by parents.[21] This information is critical to assess the feasibility of offering voluntary testing, determine the impact of such testing on clinical and developmental services, and conduct economic evaluation (e.g., cost-benefit analysis) of testing.

In this paper, we discuss formative research for and the development of a conjoint analysis survey on voluntary newborn screening in which respondents make discrete choices between two tests that are differentiated by the attributes of the diagnosed conditions and the tests.

Methods

A conjoint analysis survey is developed in several steps (Table 1). We reviewed the literature on newborn screening to develop a list of attributes likely to affect the preferences of the population of interest. We held focus groups to determine which of these attributes are most important to the population of interest and to identify any additional important attributes. Then we developed survey questions that examined the relative importance of the attributes identified as important by the literature review and the focus groups.

Identify Attributes: Literature Review

We reviewed the published literature on parental attitudes toward newborn screening to obtain the information needed to guide question and survey development, including parents' knowledge of newborn screening, their attitudes toward screening, what aspects of a genetic condition or test affected parents' attitudes, and whether knowledge or attitudes varied by the characteristics of the parents.

Table 1. Formative research and development steps to developing a conjoint analysis survey

Development Step	General Objective	Specific Objective Relative to Newborn Screening Survey
1. Identify attributes: literature review	Identify attributes of the good that may affect consumer preferences.	Identify • attributes of genetic condition, • attributes of genetic test, and • characteristics of parents that may affect parental preferences about testing.
2. Winnow attributes: expert review	Reduce list of attributes to number that can be measured effectively by a survey.	Reduce number of conditions and test attributes to seven or fewer.
3. Rank attributes: focus groups	• Confirm that participants understand attributes and their definitions. • Determine which attributes are most important. • Identify any important attributes not previously reported.	• Investigate participants' understanding of attributes to be assessed. • Determine most important attributes of genetic condition and genetic test (of seven from expert review). • Identify any important attributes not included.
4. Develop questions	Develop questions that force respondents to choose between different attributes of the good.	Develop questions that force choices between attributes of condition or test.
5. Test questions: focus groups	Confirm that draft questions are clear and correctly understood by respondents.	Confirm that draft questions are correctly understood by respondents, particularly those related to probabilities.
6. Finalize questionnaire	Finalize stated preference questionnaire.	Generate a clear, pilot-tested questionnaire.

We searched PubMed for articles with the keywords "newborn screening" and "parent," with any of the following words: "attitudes," "preferences," or "opinions." We also reviewed the bibliographies of these articles to identify relevant articles not found in the PubMed search. We excluded articles not published in English. We also excluded articles on newborn hearing screening, which requires different testing and follow-up processes and may have different implications for reproductive decision making.

We used the findings from the literature review to develop a broad list of attributes of genetic conditions and genetic tests that might affect parental decision making about newborn screening. The list was shortened in the next step of the survey development process.

Winnow Attributes: Expert Review

We narrowed the initial list of attributes from the literature review through expert review. Experts in genetics, pediatrics, newborn screening, and conjoint analysis from the Centers for Disease Control and Prevention and RTI International individually reviewed the list of attributes and ranked them in order of importance. The group then discussed the rankings. Attributes that were low ranking, that were similar to other attributes, or for which trade-offs were difficult to formulate were dropped from the list. We repeated the process until the list was reduced to seven attributes. Cost was considered to be a mandatory attribute because it allows the total value of any newborn screening test to be quantified in terms of dollars for economic analyses. Without cost data, preferences in a conjoint analysis survey can be ranked relative to one another, but they have no reference point outside of the survey.

After the ranking, we developed concise one- to two-sentence descriptions for each attribute. We also identified two to four levels of the attribute that were sufficiently different to allow clear, discrete choices between them. The attribute levels were chosen to span the relevant technology and policy options that are applicable now or that may be applicable in the near future. Characteristics of parents or of health care were included as covariates rather than as attributes for conjoint analysis.

Rank Attributes and Test Questions: Focus Groups

We conducted focused group interviews (more commonly referred to as focus groups) to test the target population's understanding of concepts before using structured data collection techniques. In a focus group, an interviewer presents structured questions and facilitates a discussion. We used focus groups to evaluate how well parents understood the attributes of interest to our study and determine which attributes were most relevant to parents of young children. We conducted two focus groups in May and June 2006 in Research Triangle Park, North Carolina. A large local market research firm recruited parents of healthy children younger than 9 months old. For families having more than one parent, we invited either parent (but not both) to participate. At least half of the participants in the second focus group were required to be from minority groups. We paid participants $75 to compensate for their time (about 2 hours) and asked them to complete an optional, brief demographic characteristic questionnaire. The project was ruled exempt by the RTI Institutional Review Board.

In the focus groups, we presented an unspecified disease for which no cure exists, although some symptoms may be treatable, and asked the participants to consider screening tests for this disease. The groups discussed topics including newborn screening in general, the conditions screened and the potential tests, and the importance of the seven attributes under consideration. Each focus group discussed the same attributes, but we used different phrasing to determine the wording participants understood the best. Focus group participants also completed risk perception exercises and two sets of example conjoint survey questions.

The first set of questions asked participants to choose between hypothetical tests for a disease given different levels of incidence, physical and mental limitations resulting from the disease, and the cost of the screening test. The second set asked participants to choose between hypothetical tests for a disease given different levels of incidence, specificity, recurrence, and the cost of the screening test. After completing each set, we asked participants to explain their choice of test. We tested their willingness to

accept trade-offs by asking what improvement in the less preferred option or what worsening of the more preferred option would cause them to switch their selection.

Results

Identify Attributes: Literature Review

Search Results

We identified 29 articles that reported primary data on the knowledge, attitudes, and preferences of parents or potential parents about newborn screening.[10,21-48] The articles reported findings in four areas: knowledge about newborn screening, attitudes toward newborn screening and factors affecting those attitudes, psychological effects of newborn screening, and the effects of positive newborn screening results on reproductive plans and actions. Study findings were consistent across countries, although they had different health care and screening programs. Therefore, we included articles from Europe and Australia, as well as from the United States.

The identified articles discuss current screening programs in general,[21,26,28,32,41] screening for cystic fibrosis,[22,24, 27,29,30,33,36,38-40,42,45,46] Duchenne/Becker muscular dystrophy,[10,14,25,29-31,37,40,44] hemoglobinopathies,[27,34] fragile X,[43] metabolic disorders,[21,25,27,30,35,40,47,48] and other diseases.[11,23,27,29,30] Thirteen studies included parents of affected children,[10,22,24,29,30,34,36,37,39,42,43,47,48] 9 included parents of healthy children with an abnormal test result,[10,21,33-35,38,45-47] and 16 included parents of healthy children or prospective parents.[10,21,23,25-30,32,40,41,44,45,47,49] The study samples were drawn from a variety of sources, including the community at large, newborn screening programs, obstetric wards and clinics, specialized clinics, support groups, and birth registries. Ten[10,24,25,28,30,34,37,39,40,45] studies used qualitative data collection methods: three studies (reported in six articles)[25-30] conducted focus groups, and seven[10,24,34,37,39,40,45] conducted narrative or semistructured interviews. Nineteen studies[10,21-24,31-33,35,36,39,41,42,44-49] used quantitative data collection: 8 used

structured interviews[21,22,32,42,44,47-49] and 11[10,23,31,33,36,39,41,43,45,46,48] used self-administered questionnaires. Some studies used more than one data collection method.

Knowledge of Newborn Screening

Our review of the literature shows that most parents are unaware of newborn screening unless their infant has had an abnormal result (Table 2).[25,28,32,40,47,49] Parents who are aware of the screening are not knowledgeable about the process of reporting screening results or the conditions for which newborns are screened.[25,28] Many parents confuse genetic screening with testing for jaundice, infections, or drug exposure.[25] Very few are aware that they could refuse testing.[30,46] Parents are unaware of and unable to consider the implications of testing, which, as we discuss in more detail below, can increase their distress if a result is abnormal. Parental knowledge of newborn screening does not appear to have improved during the 25-year span covered by this research, although the qualitative methods used make it difficult to assess trends across time.

Most parents would like more information on newborn screening[26,28,34,40] and feel that improved information would be needed even more if screening were expanded to include conditions that do not require immediate treatment.[30] Parents would prefer to receive information on newborn screening during pregnancy.[26,28,34,40,45] Most parents do not feel that their consent should be required for screening,[26,34] yet informed consent has been shown to increase parental knowledge of newborn screening during research studies.[32, 40] During routine care, however, many women sign a consent form without reading it.[28] Women who had difficult labors or whose infants had health problems found it especially difficult to comprehend the provided brochures on newborn screening just after delivery.[28,40]

Table 2. Parental knowledge of newborn screening

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population (Sample)	Condition	Knowledge of Screening	Factors Affecting Knowledge
Campbell, 2003,[25] US (Chicago), 2000	• Sample: convenience, community recruitment • Data collection: focus groups with semistructured interview	Parents (12 groups of 4–12)	PKU, sickle cell anemia	• Little knowledge of screening: - most did not know testing was done - most did not know conditions for which testing is done. - only 3 groups could name at least one screened condition; only 2 could list the health problems that caused these conditions - most were unclear about differences between genetic screening and testing for infections, drugs, jaundice, etc.	
Campbell, 2004,[26] US (Chicago), 2000	See above	Parents (13 groups)	Current practice	• Few recalled screening: zero parents (4 groups), 1 parent (5 groups), >1 parent (4 groups). • All wanted more information. Seven groups mentioned incorporation into prenatal care.	
Campbell, 2005,[27] US (Chicago), 2000	See above	Parents (12 groups of 4–12)	PKU, sickle cell anemia	• Same information as in 2003 paper.	

Table 2. Parental knowledge of newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population (Sample)	Condition	Knowledge of Screening	Factors Affecting Knowledge
Davis, 2006,[28] US (Louisiana, New Mexico, Maryland), 2003–2004	• Sample: purposeful, chosen for selected characteristics • Data collection: semistructured focus groups and individual interviews (in person or phone)	Parents of children <1 year of age (51)	Current practice	• Almost none were familiar with term "newborn screening." • Some were familiar with heel-stick test or PKU. None knew more than one condition tested for at birth. • Parents confused NBS with testing for jaundice, hepatitis B vaccination, or prenatal testing. • Many received a brochure after delivery. • Few read it or recalled the information. • Fewer recalled being told anything about NBS while in the hospital. Some recalled being told their baby had a blood test. • Most remembered signing the form. Parents did not remember information on the form. • Few knew additional screening may be available. • All felt information should be provided during third trimester prenatal care. • All preferred oral education supplemented by a written brochure. • Only needed information desired: infant would be screened, screening would benefit infant, retesting might be needed and, if so, parent would be notified.	• Language: Spanish • The Spanish phrase for NBS is easily confused with other tests done on newborns
Detmar, 2007,[30] Netherlands, 2005	• Sample: convenience, health care practices • Data collection: focus groups using a semistructured interview	Prospective parents and parents (36)	PKU	• All knew about heel stick. • Most did not know what test was for. • Only one knew parent could refuse. • Most parents did not receive any information. • A few received information but did not recall reading it.	
Detmar, 2007,[30] Netherlands, 2005	See above	Prospective parents and parents (36)	CF	• More information needed if screening expanded. • Particularly wanted information if no immediate treatment to prevent harm. • All thought information needed to be provided during pregnancy. • Some wanted to be informed early in pregnancy and some later in pregnancy.	Parental status: no differences

Holtzman, 1983,[51] US (Maryland), NS	• Sample: population, cluster sampling by hospitals
	• Data collection: structured in-person interviews
		New mothers: predisclosure (210, postdisclosure (418)	Current practice	• Disclosure statement increased knowledge of
				  – disorders for which screening is conducted
				  – effectiveness of therapies and screening test
				  – interpretation of results
				• Only 53% in predisclosure group had heard of NBS.
				• Earlier disclosure was associated with greater knowledge of NBS.
					Socioeconomic status, education, age, race, month prenatal care started

Table 2. Parental knowledge of newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population (Sample)	Condition	Knowledge of Screening	Factors Affecting Knowledge
Locock, 2008,[34] England, NS	• Sample: maximum variation, health care practices and support groups • Data collection: in-depth narrative interviews followed by specific prompts	Carriers (30), parents of carrier infants (9)	Hemoglobino-pathies (sickle cell and thalassemia)	• Antenatal carrier screening: – most thought it a routine blood test – a few thought it mandatory – most did not take test seriously or consider implications of testing • NBS: – not understood by most parents – hard to focus on information after delivery, especially if labor difficult or infant ill • Parents wanted to be more informed. It was unclear if they would prefer requiring explicit consent.	
Parsons, 2007,[40] Wales, 2001	• Sample: sequential cohort of births, midwives • Data collection: semistructured in-person interviews	New mothers (18)	PKU, congenital hypothy-roidism, CF, DMD	• Information on NBS received: – prenatal leaflet and discussion (1) – prenatal class (2) – after birth (15) • Information received verbally and from prenatal leaflet was remembered. • Difficult to read and absorb information received after birth. • Information process acceptable (4). • Information process very unsatisfactory (16): – not given enough information about NBS – literature on too many different topics – midwife did not have time to answer questions – all preferred to be informed during pregnancy • Parents saw test as routine and did not expect abnormal results. • Parents did not consider each test separately. • Parents did not recognize the diseases differed in manifestation or treatability.	
Smith, 1990,[44] Wales, NS	• Sample: sequential cohort of births, OB unit • Data collection: structured interview	New mothers (201)	DMD	• Aware of NBS (137): – primiparous (43/92) – multiparous (94/109)	Parity
Suriadi, 2004,[49] Australia, NS	• Sample: sequential cohort from NBS • Data collection: structured interview	New mothers (232)	Current practice	Aware of NBS—26%	
Tluczek, 1992,[46] US (Wisconsin), NS	• Sample: sequential cohort, NBS • Data collection: self-administered questionnaires	Parents of infants with false positives (104)	CF	• 73% aware of NBS. • 54% aware it was required by law. • 45% aware of option to refuse for religious reasons. • 29% aware could refuse CF screening for any reason.	Education

Table 2. Parental knowledge of newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population (Sample)	Condition	Knowledge of Screening	Factors Affecting Knowledge
Tluczek 2005,[45] US, (Wisconsin), 2003–2004	See above	• Families of newborns: abnormal screening results, interviews (14) • Depression scales: abnormal results (29), normal results(18)	CF	• Most only vaguely aware of NBS. • Fewer knew what tests were done. • Many learned of screening by bandage on baby's heel. • Most received an NBS brochure but did not read it. • Several wanted more information before the child was born or when blood was drawn.	

CF = cystic fibrosis; DMD = Duchenne muscular dystrophy; NBS = newborn screening; NS = not stated; OB = obstetrics; PKU = phenylketonuria.

Parental Attitudes Toward Newborn Screening

Parents and prospective parents indicate widespread support for newborn screening of conditions that require immediate treatment (Table 3).[21,26,28,30,41] Some parents also support screening for conditions that do not require immediate treatment (hereditary hemochromatosis[23] and hereditary breast and ovarian cancer[27]), for which the benefits of asymptomatic treatment are uncertain (cystic fibrosis[22,27,30,36,38,45]), or for which no treatment is available (Duchenne muscular dystrophy [10,26,30,31,37,44]). However, some parents feel that screening for untreatable conditions should be voluntary.[26,30,40]

Parents support newborn screening for conditions for which immediate treatment can be provided because it is expected to improve the infant's health care and health outcomes. Many parents support mandatory screening for such conditions, although some parents preferred voluntary screening because they feel it respects parents' religious beliefs and decision-making role. Supporters of newborn screening for disorders that do not require immediate treatment cite the following reasons:

- parental certitude about the diagnosis and avoidance of diagnostic delay

- preparation and lifestyle planning

- change in child-rearing practices

- impact on reproductive planning

- ability to teach children about condition gradually or guide child's lifestyle practices

- reduction of disease burden for society

- possibility of providing ameliorating or alternative treatments.[10,22,26,27,29,43]

The critics of newborn screening in the absence of immediate treatment cite the following reasons:

- disruption in child's identity development

- exposing of child to unnecessary interventions

- creation of uncertainty for family

- loss of carefree time before onset of symptoms

- possibility of discrimination

- creation of distress due to false positives.[22,26,27,29]

Table 3. Parental attitudes toward newborn screening

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population (Sample)	Condition	Attitude Toward Screening	Factors Affecting Knowledge
Al-Jader, 1990,[22] Wales, 1985–1989	• Sample: sequential cohort, NBS • Data collection: structured interview	Parents of children with CF (29 sets): NBS diagnosis (18 sets), clinical diagnosis (11 sets)	CF	• Support for NBS for CF: – NBS diagnosis (83%) – clinical diagnosis (91%) • Reasons for supporting screening: – improved health care and outcomes (100%) – peace of mind regarding diagnosis (16%) – reproductive decision making (8%) • Reasons for not supporting screening: – undue delay before final diagnosis (50%) – initial reservations but later support (50%) – no association with social class and attitudes toward NBS	
Bassett, 2001,[23] Australia, NS	• Sample: sequential cohort, antenatal clinic • Data collection: self-administered questionnaire	Pregnant women (135), their partners (127)	Hereditary hemochroma-tosis	• 99% would accept newborn genetic screening in general • 91.5% would accept hemochromatosis screening	None. No effect of hemochromatosis knowledge, hemochromatosis family history, ethnicity, age <30 years, tertiary education, and occupation class.
Campbell, 2003,[25] US (Chicago), 2000	• Sample: convenience, community recruitment • Data collection: focus groups with semistructured interview	Parents (13 groups)	PKU	• All supported NBS. • Mandatory (6 groups): – parents may refuse from ignorance – teenage parents were of special concern (3 groups) • Voluntary (4 groups): – respect religious beliefs – respect parental role as decision maker • Costs: – not able to breastfeed (1 group) – insurance coverage (3 groups) • Concerns: – use of blood samples for research (4 groups)	
Campbell, 2003,[25] US (Chicago), 2000	See above	Parents (13 groups)	DMD	• Support for screening: – mixed (4 groups) – supported (8 groups minus one participant) • Reasons for supporting NBS: – preparation – chance to accommodate change in child-rearing practices – lack of treatment not accepted – seek alternative treatments (8 groups) • Concerns expressed: – may be unwilling to discipline – may limit physical activities	

Table 3. Parental attitudes toward newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population (Sample)	Condition	Attitude Toward Screening	Factors Affecting Knowledge

Campbell, 2005,[27] US (Chicago), 2000	See above	Parents (12 groups of 4–12)	PKU, DMD	• All supported NBS.
				• Mandatory (6 groups):
				  – parents may refuse from ignorance
				  – teenage parents were of special concern (3 groups)
				• Voluntary (4 groups):
				  – respect religious beliefs
				  – respect parental role as decision maker
				• Costs:
				  – not able to breastfeed (1 group)
				  – insurance coverage (3 groups)
				• Concerns:
				  – use of blood samples for research (4 groups)
				• Support for screening:
				  – mixed (4 groups)
				  – supported (8 groups minus one participant)
				• Reasons for supporting NBS:
				  – preparation
				  – chance to accommodate change in child-rearing practices
				  – lack of treatment not accepted
				  – seek alternative treatments (8 groups)
				• Concerns expressed:
				  – may be unwilling to discipline
				  – may limit physical activities	

Campbell, 2005,[27] US (Chicago), 2000	See above	Parents (12 groups of 4–12)	BRCA	• Supported childhood testing for BRCA (9 of 11 groups)
				• Reasons for supporting:
				  – modify diet (6 groups)
				  – teach breast self-exam (3 groups)
				  – prepare financially (1 group)
				  – keep abreast of medical advances (2 groups)	

Campbell, 2005,[27] US (Chicago), 2000	See above	Parents (12 groups of 4–12)	CF, sickle cell anemia	• Childhood carrier testing (reproductive age or younger):
				  – widespread support
				• Reasons for support:
				  – right to know (8 groups)
				  – possible impact on sexual activity (7 groups)
				  – inform gradually or at teachable moments	

Davis, 2006,[28] US (Louisiana, New Mexico, Maryland), 2003–2004	• Sample: purposeful, chosen for selected characteristics • Data collection: semistructured focus groups and individual interviews (in person or phone)	Parents of infants (22 groups)	Current practice	Most parents felt consent was not needed. A few were concerned about cost of test or insurance coverage for test.	
Detmar, 2007,[30] Netherlands, 2005	• Sample: convenience, health care practices • Data collection: focus groups using a semistructured interview	Potential parents and parents (36)	PKU	Parents supported mandatory testing. Parents-to-be wanted a choice for each disorder.	Parental status—no differences

Table 3. Parental attitudes toward newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population (Sample)	Condition	Attitude Toward Screening	Factors Affecting Knowledge
Detmar, 2007,[30] Netherlands, 2005	See above	Potential parents and parents (36)	CF	Mixed opinions on compulsory screening or parental consent.	
Detmar, 2007,[30] Netherlands, 2005	See above	Potential parents and parents (36)	DMD	Supported optional screening (no treatment).	
Detmar, 2007,[30] Netherlands, 2005	See above	Potential parents and parents (36)	Celiac disease	Supported optional screening (increased risk only).	

Detmar, 2008,[29] Netherlands, 2005	See above	Potential parents and parents (36)	CF, DMD, celiac disease	**Reasons for supporting screening:**
				• Child
				  – reduced medical harm (all groups)
				  – avoidance of diagnostic delay
				  – lifestyle options
				• Family
				  – reproductive decision making
				  – creating certainty
				  – anticipation of future
				  – anticipated regret if not tested
				• Societal
				  – reduce disease burden for society
				Reasons against screening:
				• Child
				  – disturbed identity development (identification as sick, overprotected, spoiled)
				  – unnecessary interventions
				• Family
				  – creates uncertainty
				  – loss of carefree time
				• Societal
				  – moral consequence (blamed for additional children)
				  – discrimination
				  – false positives
				  – improved diagnostic process preferred to expanded screening	

Hildes, 1993,[31] Canada (Manitoba), 1992	• Sample: convenience, genetic counseling • Data collection: mailed self-administered questionnaire, telephone follow-up	Parents of affected child or carrier	DMD	8 of 10 supported routine screening.	
Holtzman, 1983,[32] US (Maryland), NS	• Sample: population, cluster sampling by hospitals • Data collection: structured in-person interviews	New mothers: predisclosure (210) and postdisclosure (418)	Current practice	46% preferred routine testing without consent.	Women with more knowledge of NBS preferred mandatory screening.
Locock, 2008,[34] England, NS	• Sample: maximum variation, health care practices and support groups • Data collection: in-depth narrative interviews followed by specific prompts	Carriers (30), parents of carrier infants (9)	Hemoglobino-pathies (sickle cell anemia and thalassemia)	• Most glad to know carrier status. • Carriers preferred to know before becoming pregnant or choosing a partner. • Some parties to arranged marriages felt premarital screening preferable so a different partner could be chosen.	

Table 3. Parental attitudes toward newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population (Sample)	Condition	Attitude Toward Screening	Factors Affecting Knowledge
Mischler, 1998,[36] US (Wisconsin), 1994	• Sample: sequential cohort, NBS. • Data collection: self-administered questionnaire at testing and 1 year after	Parents of children with CF (71), parents of children with false positive (106)	CF	1 year after diagnosis: 90% of parents of infants with false positives supported newborn screening.	

Parsons, 1996,[37] Wales, 1995	• Sample: sequential cohort, NBS
	• Data collection: in-person interviews
		Parents of affected child (41): communication protocol (25), clinical diagnosis or prior to protocol (16)	DMD	• Felt communication good or excellent:
				  – protocol parents (88%)
				  – nonprotocol parents (19%)
				• Average communication score:
				  – protocol (4.3)
				  – nonprotocol (2.6 [chi-square: <.05])
				• Parents commented on speed of results, time spent, and caring of doctor.
				• Supported NBS:
				  – 13 of 15 families with diagnosis by NBS
				  – 1 family with diagnosis by NBS before protocol regretted testing	

Parsons, 2002,[10] Wales, NS	• Sample: sequential cohort, NBS
	• Data collection: self-administered questionnaires, semistructured interviews, home visits (cases only)
		Parents of affected child. Diagnosis: NBS (20), clinical (16), transient increased creatine (18), healthy baby boys (43)	DMD	**Screened cohort:**
				• Supported screening (18/20)
				  – reproductive choice
				  – time to adjust to diagnosis
				  – early physiotherapy
				  – ability to plan for future
				• Undecided (1)
				• Regretted screening (1)
				  – felt information inadequate
				Transient cohort:
				• Against screening (3)
				  – did not realize DMD was untreatable (1)
				• Doubted normal follow-up test (2)
				Clinically diagnosed cohort:
				• Preferred NBS (14)
				  – plan for future
				  – early physiotherapy
				  – avoid delay in diagnosis
				  – avoid misunderstanding of symptoms
				• Against screening (1)
				• Undecided (1)
				  – would favor NBS if had second affected son	

Parsons, 2003,[39] Wales, 1999	• Sequential cohort, NBS • Data collection: self-administered questionnaires, semistructured interviews, home visits (cases only)	Parents of infant with abnormal sweat test (19), affected (9), carriers (10), other mothers (82)	CF	All 10 carrier families supported screening.	
Parsons, 2007,[40] Wales, 2001	• Sample: sequential cohort of births, midwives • Data collection: semistructured in-person interviews	New mothers (18)	PKU, congenital hypothyroidism, CF, DMD	Many felt test was routine. Others felt consent needed but did not feel they had enough information to consent. Most followed recommendation of midwife.	

Table 3. Parental attitudes toward newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population (Sample)	Condition	Attitude Toward Screening	Factors Affecting Knowledge
Prosser, 2008,[21] US (Massachusetts, Pennsylvania), 2004–2006	• Sample: population; case, metabolic clinics, and NBS; controls, birth certificates • Data collection: structured telephone interviews	Parents of children with false-positive result (66), parents of children with normal result (44)	Current practice	Parents of children with false positives were less willing than those of children with normal results to expend time or money to avoid a false positive. Both groups were willing to expend more time and money to avoid dietary treatments or developmental delay than to avoid a false-positive result.	
Quinlivan, 2006,[41] Australia, 1999–2000	• Sample: convenience, OB wards • Data collection: self-administered questionnaire	Women in public care who gave birth at tertiary care hospital (200)	Current practice	NBS was useful if • prevented a disease (85%) • reduced severity of disease (86%) • helped with reproductive planning (65%). Felt NBS: • beneficial to newborn (72%) • harmful to newborn (6%) • morally justified (63%) • against their religion (8%).	
Skinner, 2003,[43] US, NS	• Sample: convenience, fragile X study, research foundation, Web sites • Data collection: mailed self-administered questionnaire	Parents of affected children: mothers (279), fathers (163)	Fragile X	Supported testing (95%). Felt prepregnancy carrier testing was best time for testing (80%). Increased religiosity associated with stronger disapproval of testing during pregnancy.	
Smith, 1990,[44] Wales, NS	• Sample: sequential cohort of births, OB unit • Data collection: structured interview	New mothers (201)	DMD	Would accept screening for DMD (189). Would want to know at birth if child was handicapped (179).	
Tluczek, 2005,[45] US (Wisconsin), 2003–2004	• Sample: sequential cohort, NBS • Data collection: self-administered questionnaires	Families of newborns with abnormal NBS results: interviews (14). Completed depression scales: abnormal NBS (29), normal NBS (18).	CF	Supported NBS (90%). Preferred preconception carrier testing (1).	

BRCA = breast cancer gene mutations; CF = cystic fibrosis; DMD = Duchenne muscular dystrophy; NBS = newborn screening; NS = not stated; OB = obstetrics; PKU = phenylketonuria.

Other concerns raised about screening include questions about the cost of or the insurance coverage for the test,[28] the possibility of future difficulty getting insurance,[26] and the potential use of samples for future research.[26] Although some parents expressed concern that parents would be overprotective of asymptomatic diagnosed children,[26,29] others felt it was unlikely.[43]

Support for newborn screening did not vary by most parental characteristics. Parents with higher religiosity had higher disapproval of testing during pregnancy but did not differ in their support of newborn screening.[43] Parents who had distressing experiences related to newborn screening, such as a false-positive test with a long delay before diagnosis, were less likely to support newborn screening.[10,37]

Psychological Effects of Newborn Screening

Almost all parents experience some distress following notification of abnormal screening test results (Table 4). Parental reactions include shock, concern, disbelief, depression, anxiety, and confusion.[22,46] Lack of familiarity with newborn screening and delays or other issues in reporting results increased parental distress, as did delays in diagnostic testing.[22,28,34] Some parents whose children were already symptomatic were relieved to have a diagnosis.[22] Others expressed gratitude that the problem was diagnosed early.[46] In one study, a few parents temporarily rejected their infants, and other parents reported being overprotective and more attached, but these reactions were not common in other studies.[22]

Table 4. Psychological impact on parents of positive results from newborn screening

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population	Condition	Psychological Impact of Positive Result	Factors Affecting Psychological Reaction
Al-Jader, 1990,[22] Wales, 1985–1989	• Sample: sequential cohort, NBS • Data collection: structured interview	Parents of affected children (29 sets): NBS diagnosis (18 sets), clinical diagnosis (11 sets)	CF	• Experience some difficulty (100%) • Overprotective and more attached (55%) • Thought baby would die soon (28%) • Shocked (8%) • Temporarily rejected infant (14%) • Difficulty feeding infant (3%) • Relieved to know what was wrong (3%)	• Delay in diagnostic confirmation (38%) • Temporary rejection: – Social Class 3 (Social Class Classification OPCS [1972])
Boland, 1990,[24] Australia (New South Wales), NS	• Sample: convenience, CF clinics • Data collection: unstructured in-person interview; quantitative assessment tools, three subscales of Parental Attitude Research Inventory, State Trait Anxiety Inventory	Mothers of affected children: clinical diagnosis (29), NBS diagnosis symptomatic (13), NBS diagnosis asymptomatic (16)	CF	• Mothers of symptomatic NBS infants had lower scores on fostering dependency. • Mothers of asymptomatic NBS infants had higher scores on intrusiveness. • Reasons for the differences were unclear.	
Davis, 2006,[28] US (Louisiana, New Mexico, Maryland), 2003–2004	• Sample: purposeful, chosen for selected characteristics • Data collection: semistructured focus groups and individual interviews (in person or phone)	Parents of infants (22 groups)	Current practice	• Parents were unfamiliar with reporting process. – caused confusion and anxiety when contacted for retesting	

Table 4. Psychological impact on parents of positive results from newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population	Condition	Psychological Impact of Positive Result	Factors Affecting Psychological Reaction
Lewis, 2006,[33] Canada, 2002–2003	• Sample: cohort, NBS • Data collection: mailed self-administered questionnaire	Parents of carriers	CF	• Worried about child's health (29%). • Worried about health of carrier parent (3%). • Worried about effect on relationships (NS). • Worried more about CF carrier than other children (14%). • Extremely anxious when informed of need for sweat test (75%). • Remained anxious after results (20%).	
Locock, 2008,[34] England, NS	• Sample: maximum variation, health care practices and support groups • Data collection: in-depth narrative interviews followed by specific prompts	Carriers (30), parents of carrier infants (9)	Hemoglobino-pathies (sickle cell and thalassemia)	• Many did not realize they were being screened. • Often shocked that they or their infant was a carrier: – one regretted prenatal diagnosis; believed chorionic villus sampling caused infant's β-thalassemia • Informing people about carrier status by mail was distressing.	
Marsden, 2003,[35] US (New England), NS	• Sample: cohort, NBS (cases), birth certificates (controls) • Data collection: NS	Parents of children with false positives (26), parents of children with normal results (64)	Metabolic disorders	No difference in mean parental stress index.	
Mischler, 1998,[36] US (Wisconsin), 1994	• Sample: sequential cohort, NBS • Data collection: self-administered questionnaire at testing and 1 year after	Parents of children with CF (71), parents of children with false positive (106)	CF	1 year after test 7–10% of parents of children with false positives thought about test at least once a week.	
Parsons, 2002,[10] Wales, NS	• Sample: sequential cohort, NBS • Data collection: self-administered questionnaires, semistructured interviews, home visits (cases only)	Parents of affected child. Diagnosis: NBS (20), clinical (16), transient increased creatine (18), healthy baby boys (43)	DMD	• Mother–baby relationship scored higher 1 month after the report than 1 month before in transient and screened groups. • No differences on rejection or protection statements. • Mothers of healthy boys (aged 6–9 months) more likely to describe their sons as healthy, alert, great, fun, a handful, or doing well. • Mothers of NBS-diagnosed sons more likely to describe their sons as cuddly. • At 4 years old, NBS-diagnosed boys were more likely than clinically diagnosed boys to be described as responsive or great. • No differences in maternal anxiety or well-being.	

Table 4. Psychological impact on parents of positive results from newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population	Condition	Psychological Impact of Positive Result	Factors Affecting Psychological Reaction
Parsons, 2003,[39] Wales, 1999	• Sequential cohort, NBS • Data collection: self-administered questionnaires, semistructured interviews, home visits (cases only)	Parents of infants with abnormal sweat test (19), affected (9), carriers (10), other mothers (82)	CF	• No difference in mother–child relationship before and after carrier identification. • No difference in rejection index, protection index, anxiety score, or well-being. • No differences in adjectives describing the baby: – fewer affected infants described as healthy • Family concerns: – wanted other children tested; professionals reluctant – telling other family members difficult; caused tension within the family • Communication problems raised anxiety: – contact by two professionals left impression diagnosis was certain (4) – informed by general practitioner over the phone (1) – learned of diagnosis by notification of appointment with pediatrician (1) – provision of information on CF left impression diagnosis was certain	
Quinlivan, 2006,[41] Australia, 1999–2000	• Sample: convenience, OB wards • Data collection: self-administered questionnaire	Women in public care who gave birth at tertiary care hospital (200)	Current practice	Would feel guilty if baby had genetic disease (33%).	
Skinner, 2003,[43] US, NS	• Sample: convenience, fragile X study, research foundation, Web sites • Data collection: mailed self-administered questionnaire	Parents of affected children: mothers (279); fathers (163)	Fragile X	• Thought testing and early diagnosis – were unlikely to disrupt parental bonding (67%) – were unlikely (50%) or somewhat likely (41%) to endanger the baby's health – would assist in getting services (77%) – would help parents understand child's needs	
Tluczek, 1992,[46] US (Wisconsin), NS	• Sample: sequential cohort, NBS • Data collection: self-administered questionnaires	Parents of infants with false positive (104)	CF	• Parents' emotional response: – gratitude for early diagnosis (88%) – concern (98%) – shock (76%) disbelief (52%) – depression (77%) anger (48%) – confusion (61%) no reaction (4%)	

Table 4. Psychological impact on parents of positive results from newborn screening *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population	Condition	Psychological Impact of Positive Result	Factors Affecting Psychological Reaction
Tluczek, 2005,[45] US (Wisconsin), 2003–2004	See above	Families with newborns with abnormal NBS results: Completed interviews (14). Completed depression scales: abnormal NBS (29), normal NBS (18).	CF	• Shocked: – infant seemed healthy – no family history of CF – followed physician's recommendations during pregnancy • Great worry and uncertainty between NBS result and sweat test. • Parents of infants with abnormal NBS: – before sweat test: parents with false positive more depressed than parents of infants with normal NBS – after sweat test: no difference in depression score – negative correlation between time waited for the sweat test and depression score	• Prior knowledge of CF, NBS, and their carrier status reduced shock, increased or decreased worry and distress. • First-time parents worried more about health of carrier child. • Method of communicating results increased stress: – informed by phone or message – informed before sweat test could be done – informed when the physician could not discuss issues in detail. • Preferred getting information face-to-face during a routine appointment.
Waisbren, 2002,[48] US (New England), NS	• Sample: convenience, NBS, metabolic clinics • Data collection: structured telephone interviews and mail questionnaires	Parents of affected children: diagnosed by NBS (28), clinically diagnosed (17)	Homocystinuria, galactosemia, maple syrup urine disease, biotinidase deficiency	No difference in stress scores between cohorts.	
Waisbren, 2003,[47] US (Massachusetts, Pennsylvania, Maine), NS	• Sample: convenience, NBS, metabolic clinics, controls, birth certificates • Data collection: in-person structured interviews (affected), structured telephone interview (false-positive or normal)	Parents of children: diagnosed by NBS (50), clinically diagnosed (33), with false positive (94), with normal results (81)	Metabolic disorders	False-positive group: • experienced more parental stress • exhibited more dysfunction in parent–child relationship	• Lower stress and dysfunction among false-positive group. • Informed face-to-face. • Referred to a metabolic center.

CF = cystic fibrosis; DMD = Duchenne muscular dystrophy; NBS = newborn screening; NS = not stated; OB = obstetrics.

The distress and anxiety felt by most parents of children with false-positive results resolved after diagnostic testing was completed.[10,33,36,45] A minority of parents continued to think about the test results,[36] worry about the child's health, or remain anxious.[33] One study found that parents of children with false-positive results were more stressed and their relationship with their child more dysfunctional than parents who had children with normal test results.[47] Most studies, however, found little long-term difference in parental depression,[45] anxiety, well-being, or rejection or protection between parents of children with false-positive results and those of children with normal results.[38] Parents' stress was reduced when they received better information.[37,45,47]

Effect of Positive Newborn Screening Result on Reproductive Plans and Actions

Some parents report that they changed their reproductive plans following the diagnosis by newborn screening of a child affected by cystic fibrosis or Duchenne muscular dystrophy (Table 5).[10,42] They decided against having additional children, planned more or fewer children, or delayed their next pregnancy.[10,42] There was little concordance between parents' plans following the diagnosis and their subsequent actions. The majority of parents who reported that they did not want more children later decided to have children, and the majority of those who reported that they wanted a larger family size than they initially planned later decided to have no more children. For parents of children with cystic fibrosis, reproductive actions were related to the health of the affected child—parents were more likely to have additional children if the child was in relatively good health and the family was coping well.[42]

At the time of diagnosis, over half of the parents expected to use prenatal diagnosis for future pregnancies.[10,22,42] Expectations about termination of the pregnancy varied and were affected by the religious views of the participants.[10,34] The use of prenatal diagnosis differed from the initial intentions for 67 percent of parents, and changes occurred in either direction.[42] In the two studies that examined outcomes of pregnancies subsequent to diagnosis, all nine affected pregnancies were terminated.[10,42]

Table 5. Effect of positive newborn screening result on parental reproductive plans and actions

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population	Condition	Effect on Future Childbearing Decisions	Factors Affecting Relationship to Reproductive Decision Making

Al-Jader, 1990,[22] Wales, 1985–1989	• Sample: sequential cohort, NBS
	• Data collection: structured interview
		Parents of affected children (29 sets): NBS diagnosis (18 sets), clinical diagnosis (11 sets)	CF	Plan more children (69%).
				Antenatal diagnosis: yes (55%), no (25%), uncertain (20%).
				Attitude toward termination:
				• Parents of screened infants:
				  – would terminate (61%)
				  – would not terminate (11%)
				  – uncertain (27%)
				• Parents of clinically diagnosed infants:
				  – would terminate (36%)
				  – would not terminate (45%)
				  – uncertain (18%)
					Social class: no difference
					Attitudes toward termination: no difference

Lewis, 2006,[33] Canada, 2002–2003	• Sample: sequential cohort, NBS • Data collection: mailed self-administered questionnaire	Parents of carriers	CF	No effect on reproductive decisions (82%). Decided to have no or fewer children (18%).	
Locock, 2008,[34] England, NS	• Sample: maximum variation, health care practices and support groups • Data collection: in-depth narrative interviews followed by specific prompts	Carriers (30), parents of carrier infants (9)	Hemoglobino-pathies (sickle cell and thalassemia)	Felt early screening and diagnosis were important. Easier to terminate earlier in the pregnancy.	Religious faith

Table 5. Effect of positive newborn screening result on parental reproductive plans and actions *(continued)*

First Author, Publication Date, Country, Study Year(s)	Study Methods	Population	Condition	Effect on Future Childbearing Decisions	Factors Affecting Relationship to Reproductive Decision Making
Mischler, 1998,[36] US (Wisconsin), 1994	• Sample: sequential cohort, NBS • Data collection: self-administered questionnaire at testing and 1 year after	Parents of children with CF (71), parents of children with false positive (106)	CF	Knew prenatal diagnosis was available (84%). Prenatal diagnosis for subsequent pregnancy: 1 of 12 had prenatal diagnosis.	
Parsons, 2002,[10] Wales, NS	• Sample: sequential cohort, NBS • Data collection: self-administered questionnaires, semistructured interviews, home visits (cases only)	Parents of affected child. Diagnosis: NBS (20), clinical (16), transient increased creatine (18), healthy baby boys (43)	DMD	Changed their reproductive plans (16): • no future children (4) • delayed their next child (11). Mean interpregnancy interval: • healthy cohort (29 months) • NBS-diagnosed cohort (41 months). Prenatal testing (19 of 27 pregnancies). Elective terminations (4).	
Sawyer, 2006,[42] Australia, 1997 and 2002	• Sample: convenience, CF clinic • Data collection: Structured in-person (preferred) or telephone interview	Parents of NBS-diagnosed children (56)	CF	Changed planned family size (19): • more children (6): – decided against more later (4) • fewer children (13). No change in planned family size (33). Did not plan family size (4). Did not want more children (27): • later wanted more children (16) Planned prenatal diagnosis (42 [82%]): • to prepare (16) • to decide about termination (18) • to terminate (12). Would not use prenatal diagnosis (9). Changed views on prenatal diagnosis (25). Pregnancies since diagnosis: • 26 women had 55 pregnancies • 67% used prenatal diagnosis for ≤1 pregnancy • one-third planned to terminate if CF • 5 of 5 affected pregnancies terminated. Hypothetical same as actual (67%). Changes in both directions.	• Coping • Health of child with CF
Skinner, 2003,[43] US, NS	• Sample: convenience, fragile X study, research foundation, Web sites • Data collection: mailed self-administered questionnaire	Parents of affected children: mothers (279), fathers (163)	Fragile X	Most thought testing and early diagnosis would inform reproductive planning (77%) and would inform family about risk of being carrier.	
Smith, 1990,[44] Wales, NS	• Sample: sequential cohort of births, OB unit • Data collection: structured interview	New mothers (201)	DMD	Definitely or probably terminate an affected pregnancy (142).	

CF = cystic fibrosis; DMD = Duchenne muscular dystrophy; NBS = newborn screening; NS = not stated; OB = obstetrics.

Attributes

In the studies we reviewed, we identified 26 features of a genetic condition or screening test related to parental decision making regarding newborn screening (Table 6). We developed a list of these features and levels of each feature to be considered for inclusion in the conjoint analysis survey.

Table 6. Potential attributes and disposition at each step of winnowing process

Attribute of Condition	Ranking by Expert Review 1	Ranking by Expert Review 2	Attribute Considered by Focus Groups	Ranking by Focus Group 1	Ranking by Focus Group 2	Volunteered by Focus Groups
Developmental disability	3	1	Yes	1	1	
Physical disability	4	2	Yes	4	3	
Frequency of acute symptoms	8	Not ranked	No			
Lifespan (years)	1	5	Yes	6	4	
Recurrence (risk in siblings)	6	4	Yes	2	5	
Incidence (risk in population)	5	2	Yes	5	2	
Perceived risk status	14	Not ranked	No			
Risk to grandchildren	Not ranked	Not ranked	No			
Age symptoms begin	2	9	No			
Diagnostic delay: number of doctor visits or time between symptom onset and diagnosis	12	7	No			
Sex of affected children	Not ranked	Not ranked	No			
Extent of treatment	6	Not ranked	No			
Availability of treatment	18	Not ranked	No			
Complexity of treatment	15	Not ranked	No			
Painfulness of treatment	17	Not ranked	No			
Duration or frequency of treatment	16	Not ranked	No			
Effectiveness of treatment	9	Not ranked	No			
Degree of disability	10	Not ranked	No			
Type of disability	11	Not ranked	No			
Variability in phenotype			No			Yes
Attribute of Test	**Ranking by Expert Review 1**	**Ranking by Expert Review 2**	**Attribute Considered by Focus Groups**	**Ranking by Focus Group 1**	**Ranking by Focus Group 2**	**Volunteered by Focus Groups**
Out-of-pocket cost	1	8	Yes	7	7	No
Number of samples required	7	Not ranked	No			No
Type of samples required	2	Not ranked	No			No
Follow-up testing	5	Not ranked	No			Yes
Sensitivity	4	9	No			Yes
Specificity	3	6	Yes	3	6	No
Positive predictive value	Not ranked	Not ranked	No			No
Negative predictive value	Not ranked	Not ranked	No			No
Timing	6	11	No			No
Precision of test			No			Yes
Invasiveness of follow-up testing			No			Yes
Side effects of follow-up testing			No			Yes

Winnow Attributes: Expert Review

Seven experts reviewed the 26 attributes identified from the literature review and the suggested attribute levels and ranked them by priority for inclusion. The lowest-ranking attributes after the first round of expert review were frequency of acute symptoms, perceived risk status, risk to grandchildren, age symptoms begin, and the sex of affected children.

The review group then met, discussed the full list of attributes and the rankings, and further narrowed the list to 11 items: mental (developmental) disability, physical disability, lifespan, recurrence risk in siblings, incidence (risk in newborn population), age at which symptoms begin, the average time or number of doctor visits from onset to diagnosis, the out-of-pocket cost for the test, the sensitivity of the test, the specificity of the test, and when the test is done.

This shortened list was then ranked by an expanded list of experts. After the second ranking, the six highest-ranked attributes were developmental disability, incidence, lifespan, physical disability, recurrence, and specificity (false-positive rate). The length of time between onset of symptoms and diagnosis, often referred to as the "diagnostic odyssey," was ranked seventh, and the cost of the test was ranked eighth. Because cost was a required attribute, diagnostic odyssey was dropped. Thus, the attributes to be considered by the focus groups were cost, developmental disability, incidence, lifespan, physical disability, recurrence, and specificity (false-positive rate). The descriptions and levels of the attributes presented to the second focus group are shown in Table 7.

Rank Attributes: Focus Groups

We conducted two focus groups. Each had nine parents of infants, for a total of 18 participants: 10 women and 8 men. All participants completed the optional demographic questionnaire. Eleven participants identified themselves as non-Hispanic Caucasians and seven as non-Hispanic African American. The mean age of the participants was approximately 29 years, and the mean age of the infants was about 5 months. All participants were high school graduates, and 11 participants had 4 or more years of college. Fifteen participants were employed full-time, two were full-time parents, and one was unemployed and seeking work. The median household income for all participants was about $63,000 per year, slightly higher than the average in the Raleigh-Durham, North Carolina, area in 2006. Although we attempted to recruit a diverse group of participants for both focus groups, the first focus group included only non-Hispanic Caucasian people and had higher educational and income levels than the second group.

Knowledge of Newborn Screening

To identify what background information would be needed in the survey, we asked focus group participants what they knew about newborn screening. Participants in both groups had minimal knowledge of newborn screening. Although they were aware that their newborns had been tested, none knew the specific conditions for which their infants had been tested. Two parents learned of newborn screening tests from either childbirth preparation class (one parent) or a prenatal pediatric visit (one parent). Others said that their prenatal care provider had told them about the screening tests, but it was unclear whether parents distinguished newborn screening from prenatal screening. None of the parents were aware that they could opt out of newborn screening testing or that additional screening tests were available independent of the state-mandated screening. As mentioned above, some participants confused newborn screening with prenatal screening, such as the triple screen for Down syndrome and neural tube defects. Several parents explained that they had some experience with these prenatal tests and noted the negative consequences of false-positive results experienced by themselves, family members, or friends.

Discussion of Attributes

Parents understood the attributes on the list and found them relevant to decision making, although some were more relevant than others. Developmental disability was ranked as the most important attribute, and the cost of the test was ranked as least important (Table 6). As reflected in the rankings, parents were more concerned about mental limitations (developmental disabilities) than they were about physical limitations or disabilities. The degree

Table 7. Attribute descriptions and levels

Attribute	Description	Levels
Cost	Your cost is the money that you would personally have to pay. It is your share of the fees that are not covered by your insurance or health care plan.	• $500 • $300 • $150 • $75 • $25 • No charge (free or completely covered by insurance)
Mental limitations	Genetic health problems may involve mental limitations that limit a person's ability to do various activities of daily life, such as speaking, walking, dressing, eating, bathing, learning, working, and getting along in social situations and school activities.	• Level 1: – can learn at the 6th-grade level – can live and work in many jobs as an adult with some assistance – can learn to speak clearly and take care of personal needs • Level 2: – can learn at the 2nd-grade level – can live and work at simple jobs as an adult with moderate assistance – can usually overcome difficulties with daily activities such as speaking clearly and taking care of personal needs – has to live in a setting that provides some care for special needs • Level 3: – can eventually learn to take care of personal needs, but much later than other children – can learn to speak, but limited to basic words – has to live in a setting that provides medical and nursing care
Chance that a baby will have a problem	The chance that a baby will have a problem indicates how likely it is that a new baby will have a problem. The chance is the number of babies that have the problem out of 10,000 births (10,000 is about the number of people living in a small town).	• 50 out of 10,000 births • 25 out of 10,000 births • 10 out of 10,000 births • 5 out of 10,000 births • 1 out of 10,000 births • Less than 1 out of 10,000 births
Average age at death	Average age at death refers to how old someone will live to be on average. A genetic problem may reduce the average age at death relative to someone without a problem.	• Infancy (<1 year of age) • Early childhood (<10 years of age) • Adolescence (14–16 years of age) • Early adulthood (20–30 years of age) • Adulthood (40–50 years of age) • Normal (77.5 years of age)
Physical limitations	Physical limitations affect the ability to move. These limitations vary and can affect general muscle coordination, ability to walk, bladder control, etc.	• Level 1: – limits on daily physical activities such as walking or standing – can move with help from a cane, wheelchair, or other equipment – sometimes needs a little help from other people • Level 2: – greater limitations on daily physical activities – can be partly helped by using equipment – frequently needs some help from other people • Level 3: – complete loss of ability to do physical activities without help – cannot be helped by using equipment – always needs help; must live in a setting that provides extensive care
Chance that additional babies born to the same parents will also have a problem	How likely it is that later brothers or sisters of a child with this problem will have the same problem.	• 50 out of 100 births • 25 out of 100 births • 10 out of 100 births • 5 out of 100 births • 1 out of 100 births
Chance of a false-positive test result	A false-positive test result occurs when the test indicates there is a problem, but the baby actually doesn't have a problem. A positive test result usually has to be confirmed with additional or repeated tests to make sure there actually is a problem.	• 100 out of 1,000 tests • 50 out of 1,000 tests • 20 out of 1,000 tests • 10 out of 1,000 tests • 1 out of 1,000 tests

of disability was also important; parents seemed very concerned about whether the developmental disability attribute was "severe." Parents were also concerned about the amount of uncertainty in the prognosis for a condition. They distinguished between conditions for which the prognosis is very specific and predictable (i.e., all affected children are severely disabled), versus those that have a less predictable prognosis (i.e., some affected children are mildly disabled, but others are severely disabled). Participants had difficulty differentiating between the attributes in isolation (e.g., developmental disability vs. cost of test) without first imposing arbitrary levels on the attributes (e.g., level 1 developmental disability vs. $75 cost of test). In an actual survey, the levels are specified in the conjoint analysis trade-off questions, so this difficulty would not exist.

We asked participants if additional attributes of conditions or tests could affect their decision about optional newborn screening. Participants generally thought that the seven attributes presented captured the factors most relevant to decision making. When pressed, the participants identified the following additional attributes: sensitivity, uncertainty about prognosis, invasiveness and risk of diagnostic testing (if required following a positive screening test), and difficulty with obtaining health insurance after diagnosis. In discussion, many participants found the prospect of a false negative more worrisome than a false positive. Additionally, all respondents grasped the concept of sensitivity, or false negatives, but some seemed confused by false positives, even after discussion. Thus, after reviewing the attribute rankings and the additional ones volunteered in discussion, we made one significant change by replacing specificity (false positive) with sensitivity (false negative). The final attributes selected for the questionnaire were developmental disability, physical disability, incidence, recurrence, lifespan, sensitivity, and cost.

Develop Questions

The next step in the survey development process was to convert the attributes and levels identified into choice questions suitable for the intended respondents. For conjoint analysis surveys, this step involves applying statistical principles in the theory

of experimental design[53] with practical applications of psychometrics.[54,55] For space considerations, we do not present further detail on the question development process and refer the reader to these references for further information.

Test Sample Preference Questions: Focus Groups

At this stage of survey development, our goal was to see if respondents could complete sample preference questions and if the trade-offs presented to them would be salient and meaningful, determined by both variation in their responses and by informal discussion in the focus groups. To assess this, participants completed two sets of example conjoint analysis survey questions (similar to Figure 1 on the following page), each of which offered a choice between two test scenarios described with different levels of the attributes. The attribute-level ranges and combinations tested produced good variation in responses, as desired and statistically necessary in practice.[53] For one question, the nine responses were evenly split between test A, test B, and neither test, with no respondents choosing "don't know." For the other question, two respondents chose one test, six chose the other test, three chose neither, and two chose "don't know." Actual responses in a full fielding are determined jointly by respondent preferences and experimental design or the combination of attribute levels and choices presented.[53]

Participants were able to understand the questions under consideration fairly well. Most participants could articulate their reasons for their choices and were willing to accept reasonable changes in test features that would cause them to switch their selection. Four of the attributes examined—incidence, recurrence, lifespan, and specificity—are probabilistic concepts, which some parents found challenging. This challenge was largely overcome by presenting the risk level verbally as the number of cases in 1,000 births and graphically by showing colored squares in a grid of 1,000 squares to provide alternative representations of the same risk levels (Figure 1). For conditions that were indicated to be especially rare, some subjects said that they would not choose either test at the costs indicated.

Figure 1. Presentation of probabilities

	Test A		Test B	
Chance that a baby will have a genetic problem		1 out of 1,000 babies (0.1%)		20 out of 1,000 babies (2%)
Chance of a false-positive test result		20 out of 1,000 tests (2%)		50 out of 1,000 tests (5%)
Chance that later brothers or sisters will also have the genetic problem		5 out of 100 babies (5%)		25 out of 100 babies (25%)
Cost of test	**$75**		**$300**	
I would choose Test A	☐			
I would choose Test B	☐			
I wouldn't choose either test	☐			
I don't know what I would choose	☐			

Individual personality affected whether participants would choose testing. Self-described planners wanted as much information as possible for long-term decision making. For example, they wanted to know if they would have to alter their retirement or dependent care plans. Those who did not want the information did not want to mar time with their child with prior knowledge of the child's diagnosis before he or she developed symptoms. One parent supported testing because she anticipated a long delay between the onset of initial symptoms and subsequent diagnosis because "I know doctors don't listen."

Finalize Questionnaire

After the focus groups, we reviewed all survey materials tested, revised them as appropriate, and finalized them to create the full survey questionnaire. The last step in survey development was to hold a series of one-on-one pretest interviews to evaluate materials and finalize the questionnaire. We made minor revisions to layout, wording, and ordering between these interviews, but we did not make any major changes to the conjoint analysis design.

Discussion

The literature contains surprisingly little information regarding parental attitudes toward newborn screening, but some common themes did emerge from the review. Parents know very little about newborn screening and feel their level of knowledge is inadequate. Many prefer receiving information about newborn screening during pregnancy rather than after delivery, so that they can consider the information at their leisure. Virtually all parents support newborn screening for conditions requiring early treatment. Many parents also support newborn screening for conditions for which early treatment is not needed or helpful, but there is more variation in attitudes about screening for these conditions. The support for screening for untreatable conditions may result in part from unrealistic expectations of early diagnosis to improve outcomes, even in the absence of a treatment.

Our focus group findings were consistent with the literature: The parents in our groups had minimal knowledge of newborn screening, and most of the attributes identified from the literature had relevance with the parents in our groups. Parents in our focus groups also were concerned about the variability in the phenotype of the condition, the attributes of follow-up testing, and possible problems with insurability.

Existing studies on newborn screening, including this one, are limited by small sample sizes. Some studies also are limited by an inappropriate or no comparison group and by inconsistent methodologies. Often, studies used different sampling and data collection methodologies for parents of affected children or those with false-positive results and parents of children with normal screening results, which could bias results. The data collected by structured questionnaires are limited by the questions asked, so unexpected attitudes or opinions may not be captured.

Focus groups yield data that are, in essence, formative in nature. Therefore, it is not appropriate to rely solely on their data for definitive hypothesis testing. Focus groups rely on all participants' providing honest responses. In some cases where questions may have a

strong social bias, respondents may be biased toward providing responses they think are "appropriate." Although we do not think this occurred during the focus groups described in this study, it is a potential limitation for all focus group research. Likewise, given that there is wide variation in various cultures' acceptance and understanding of developmental disorders, one might anticipate that responses might vary among different cultures.

The focus groups and questionnaire pretesting provided valuable information about the development of the conjoint analysis questionnaire. The attributes of the conditions and tests that were important to parental decision making differed from those identified by the expert review panel, most notably on the importance of specificity versus sensitivity.

The conjoint analysis exercises in our draft questionnaire did not place an unacceptable burden on our focus group participants. We split the attribute list into a two-part task so that attributes of the condition and attributes of the test did not have to be evaluated simultaneously. This reduced the cognitive burden of the trade-off tasks, but it is unclear if varying all seven attributes simultaneously would be as acceptable.

The focus groups identified the importance of addressing the consistency of the prognosis in the definition of the disability attribute levels or in the adjunct information. The focus groups also identified that refusing a test was an important option for some conditions; some participants would not chose to test for rare conditions regardless of the test characteristics. This decision is very important in the context of voluntary newborn screening.

The variation in the responses to the questions suggests that the identified attribute levels are salient to respondents and cover a sufficient range to identify statistically the important factors guiding parental preference. Several factors influence optimal variation in conjoint analysis studies, but, generally, 65 to 75 percent of the sample should prefer one choice over another.[56]

Many of the attributes relevant to newborn screening concern probabilistic information. Although considerable research has shown that

even highly educated researchers have difficulty completing problems involving varying levels of risk or uncertainty,[57,58] this survey was designed to be completed independently. Providing sample or "quiz" questions to respondents before the actual survey questions may allow respondents to become acquainted with the exercises, ease respondent burden, increase data reliability, and provide comparative tests of internal validity.[3,55]

Our review of the literature and findings from the focus groups highlight the need to provide parents with information on newborn screening during pregnancy, rather than after birth. Other studies demonstrate the need for an efficient, sensitive protocol for communicating results and providing follow-up counseling and testing.[37,45,47] Delayed follow-up testing and poorly communicated test results generate increased parental stress and reduce their support for screening. Without well-designed procedures, voluntary screening programs could produce unwarranted stress and anxiety.[37] It is critical to ensure that parents do not have poor experiences with voluntary programs, which could reduce their support for screening that is important to their infant's health; the literature and our focus groups show that parents do not differentiate between different types of prenatal and newborn screening.

Finally, the conjoint analysis survey we developed provides an opportunity to contribute data on both parent preferences and the economic value of testing, which is not well established.[20] Preference data help the public health and clinician communities provide the information and the choices that parents want.[2] Our findings and those of other researchers suggest that the positive and negative characteristics that parents consider important in decision making can differ from those that clinicians or policy makers consider important.[25] The conjoint analysis survey also provides an opportunity to use the preference data to conduct a formal economic evaluation of screening, such as cost-benefit analysis, and to estimate uptake rates. The data could formally estimate who may select screening and what health

problems are of the most concern. This information could be used to prioritize test development and the conditions included in public newborn screening programs, for which resources are likely to be limited, and to develop educational materials for parents and physicians.

Future Directions

The 2002 Institute of Medicine report *The Future of the Public's Health in the 21st Century*[59] promoted expanded involvement of communities in public health research and policy decisions, recommending

> Government and private funders of community health initiatives should focus on long-lasting change by supporting ongoing community engagement and leadership through supportive mechanisms and realistic expectations.[59,p.5]

Stated preference surveys, including conjoint analysis, can provide valuable information to public health agencies on the importance of different attributes of a program or service to community members and the economic value they place on the program or service under different scenarios.[7] We can expect a growing call for quantitative data on costs and benefits in an era of resource constraints and health care reform. Conjoint analysis provides one such way to help inform policy makers about parents' views of the costs and benefits of these programs while also providing the public health and advocacy communities with information on parental preferences regarding specific expanded and voluntary newborn screening features. For example, a conjoint analysis study of Canadian parents of young children with hearing loss found that parents preferred less expensive options for care, favoring clinic-based services over home-based services and weekly visits over more frequent ones.[60] They also valued well-coordinated care with access to support from other parents, however.[60] Implementation of the survey described in this manuscript could lead to new studies of newborn screening options that would help tailor program design and prevent investment in underused public health programs.

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