

# The New England Genetics Workforce Project

## New England Region 1 Interview Pilot

### Phase One Report

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Conducted by the New England Regional Genetics Collaborative at the University of New Hampshire for the National Coordinating Center for the Regional Genetic and Newborn Screening Services Collaboratives (NCC)

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## Executive Summary:

This report reflects the findings of a study conducted in 2009-2010 on the genetic providers in the northeast providing services to patients who screen positive for a metabolic condition. The study examined both the newborn screening process in five of the six New England states (Massachusetts did not participate), and then examined through key informant interviews, the care process for a most-seen genetic patient through the first year of life. We specifically asked about the range of care, the length of visits, lab tests ordered and follow up with providers and families. We then asked about the complexities that hinder a coordinated care process, as well as best practices to dealing with those complexities. Here we summarize the findings of the study and offer insights into what we believe are the critical touch points affecting the current and future genetic workforce in the states studied both currently and in the near future.

### *Newborn Screening:*

We found that, overall, the newborn screening functions work well in the states studied. The processes did, however, differ by state. This was especially true with long-term follow-up, although these disparities are currently being discussed and addressed. Where processes do diverge, opportunity may exist for alignment.

### *Care process:*

Several themes were identified. However, it is important to note that the care process for genetic conditions can vary widely depending on the condition. Many are highly medically intensive following positive screen while others can be treated dietetically without hospitalization. Still other conditions are so rare that many providers may never see a patient with the condition. This diversity of both time and knowledge creates issues related to the themes outlined here and throughout this report. The following are three major themes suggested by findings.

1. Reimbursement is lacking

Other studies have found that reimbursement rates for genetic services are poor<sup>1,2</sup> and that medical geneticists report low satisfaction with income and earning potential.<sup>2</sup> Here that lack of reimbursement was reiterated. This includes both care time, but also a heavy administrative burden. One nutritionist estimated that they spent nearly 60 hours of administrative work per case in the first year of life. This is largely non-reimbursable. Currently little information exists on the most cost-effective methods for treating patients with metabolic conditions. More empirical evidence needs to be developed relative to the cost effectiveness of treatment and the development of best practices.

2. The need for improved care coordination.

These findings suggest that there exists a need for improved case management for metabolic patients. Those interviewed indicated that while conditions are rare, many are time intensive. And, because they are rare, it is often unclear what the correct flow of contact and care should be. This also differed by state, and respondent. Findings suggest that, new models should be explored that incorporate different roles for and/or team members for dealing with at least some patients. Most respondents cited team members as including a geneticist, nutritionist, and often nurse depending on condition. However, very few mentioned genetic counselors, psychologists, nurse practitioners, primary care physicians, or NBS coordinators as team members. Specific action protocols should also be explored to better direct time and efforts effectively. Both of

these fit well with the foundations of the medical home model that has informed health care reform and the role of accountable care organizations.

3. The education component is diverse, uncoordinated and time consuming

Education is a primary function of dealing with metabolic patients, their families, and often times their primary care providers. Currently, who engages with these various entities and how differs widely in practice. Families also have a wide range of information sources that do not provide consistent information, and not always empirically supported. These issues will no doubt exacerbate as with direct to consumer testing and enhanced screening become more prevalent.

Providers are also unclear as to their roles relative to metabolic patients. Some appreciate an active role, while others assume the patient simply transfers to another care system. Coordination of care with the PCP and the education surrounding the patients is also an important consideration that currently varies widely in practice. Care “team” members, sometimes virtual and sometimes physical, often did not know the specifics of other parts of the care process, for example, how parents may obtain dietetic formula, or what happens when there is a need during off hours or on weekends.

Findings suggest that attention be directed at developing care protocols for some if not all conditions. Also, a more focused care coordination role should be assumed. There should be conducted a review and synthesis of educational tools and best practices for families as well as a providers.

The field of genetic medicine is not currently well understood, both within and beyond the medical care system. There is no reason to think that expansions in screening or advances in the field will be any more so. The field is also approaching a potential shortage of providers that needs to be addressed. If it is not, even moderate gains in effective and efficient care as outlined here will be difficult to attain. This is especially true as the field continues to expand its knowledge and practice base. This study provides at least some empirical evidence that the current supply of genetics providers is ill equipped to accommodate future growth. Benchmarking current supply of providers relative to demand is required to better anticipate future growth needs as the field changes and expands its services and scope.

Finally, the field should take an ‘outside in’ rather than ‘inside out’ perspective. That is to say that currently the field pushes information to patients and providers hoping that the medical providers, insurers, families and states will react accordingly to address the needs of patients and the field itself. And while this makes sense in some areas, it does not in all. For example, with reimbursement, the field should be focused on where the health system as a whole is moving, how care is valued and reimbursed, and how to provide that information. Similarly it may also require rethinking care teams, their make up and roles, and the nature of care coordination and a medical home for conditions that are rare and hard to manage at times.

**Overview:**

This study proposed to conduct an assessment of the resources needed to provide quality genetic health care to an expanded population of metabolic disorder patients identified by increased newborn screening efforts. These resources include, but may not be limited to laboratory, public health, primary care, geneticists, genetic counseling personnel, equipment and services. The authors hypothesize that these resources are affected by the number of screened conditions, the type of screened conditions, demographic changes, workforce changes, and social factors. The study, conducted in phases, proposes to examine known or estimated prevalence rates by metabolic condition screened. While many studies have reviewed the health care workforce in relation to overall demographic trends, newborn screening is unique in that it is a service mandated for use by the entire population of infants. The issue is not estimating utilization, but defining and estimating resource use and allocation and the resulting infrastructure demands caused by the expanded scope of testing and identification of rare diseases not previously screened. These become especially salient later in the life course of many disorders.

**Background and Rationale:**

Newborn screening is universally mandated for infants born in the United States and territories in order to test for metabolic or other genetic disorders not otherwise apparent at birth. Without early detection, intervention and treatment, many of these disorders are fatal or cause significant medical and developmental complications. Newborn screening is a state-based core public health function, allowing affected children to be treated before clinical symptoms present, improving or mitigating adverse health outcomes. In 2006, the American College of Medical Genetics released a report outlining a standardized process for identifying and recommending conditions appropriate for screening in the newborn period.<sup>3,4</sup> The findings identified 29 conditions for which screening should be mandated, although the implementation of this recommendation varies from state to state. These conditions are a combination of endocrine, metabolic, hemoglobin, vitamin and other disorders (e.g., cystic fibrosis, congenital hearing loss). The report also lists an additional 25 “secondary target” or report-only conditions which are not actively sought by newborn screening because they do not have documented treatments or there is limited knowledge of their natural history, although they are often revealed in the course of newborn screening for the 29 core conditions.<sup>3,4</sup>

The Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) has developed a process to nominate and review conditions for addition to the Committee’s recommended uniform screening panel. Since the initial panel was defined, 9 additional conditions- Krabbe Disease, Severe Combined Immunodeficiency (SCID), Pompe Disease, Fabry Disease, Niemann-Pick Disease, Spinal Muscular Atrophy, Hemoglobin H Disease, Hyperbilirubinemia/Kernicterus, and Critical Cyanotic Congenital Heart Disease- have been considered for inclusion on the NBS panel.<sup>5</sup> Only SCID has since been added to the panel,<sup>6</sup>; SACHDNC recently recommended Critical Cyanotic Congenital Heart Disease for addition to the NBS panel to the Secretary of Health and Human Services.<sup>7</sup> It is inevitable that as laboratory technology and clinical genetics advance, newborn screening will expand to include a wider range of diseases.

Improvement and expansion of newborn screening, while benefiting population health overall, contains costs associated with implementation including increased levels of state resources such as equipment and personnel for testing and long-term follow-up of affected patients and families. The ACMG report identified that

“the public health system faces many challenges as newborn screening capabilities continue to evolve. The health care service infrastructure is limited with regard to the interconnections among primary care professionals and subspecialists, particularly in rural areas—a problem complicated by the number and diversity of very rare conditions identified in newborn screening programs”.<sup>4</sup>

The economic downturn has had a serious impact on the state-based public health workforce. Between July 2008 and January of 2009, state public health agencies lost 1,500 jobs through layoffs and attrition.<sup>8</sup> In fiscal year 2010, states expect to lose an additional 2,600 jobs funding cuts escalate.<sup>8</sup>

Newborn screening, while expanding in scope, is only one program that impacts the genetics workforce. A 2005 survey found that the medical geneticist workforce does not appear sufficient to meet the expected patient care needs for clinical genetics services in the next 5-15 years.<sup>2</sup> The integration of genetics into all areas of medicine in general comes at a time when the number of physicians choosing to practice the primary specialty of medical genetics is declining.<sup>9</sup>

### **Methodology:**

Phase one of this project sought to define the care process as it currently occurs for metabolic patients in the northeast region from initial screen to treatment at one year given a positive screen. Metabolic conditions were selected because they provide a more definable treatment protocol than some other disorders and thus allow for more accurate empirical measurement of the resources needed to treat those patients. States were used as the unit of analysis due to the unique screening process inherent in each.

During phase one, the project convened an expert advisory group to develop a research strategy to fully capture all care resources used. These were comprised of members of the New England Genetics Collaborative (NEGC), the American College of Medical Genetics (ACMG), and others. The research team was comprised of Robert McGrath, principal investigator in the Department of Health Management and Policy at the University of New Hampshire, Monica McClain, project manager for the NEGC at the University of New Hampshire, and Michelle Stransky, research assistant and doctoral candidate at the University of New Hampshire, Department of Sociology. Input on research methodology and development of the interview protocol was provided by Judith Benkendorf and Meredith Weaver at the ACMG.

In-depth interviews were conducted within each state in NEGC with both NBS coordinators to assess the NBS process in each state, and with metabolic care providers. Invitations to participate in the project were sent to the 14 metabolic clinicians in the region and to the 6 NBS coordinators in New England. Eight clinicians consented, and seven completed interviews. Five NBS coordinators consented and all completed interviews. (One declined because they do not do the referral/follow-up; the NBS lab provides these services.) The clinicians then referred members of their team to us (psychologist, dieticians, etc). We received a total of nine referrals, seven of whom completed the informed consent. Of these seven, five completed interviews. Interview guides for both the NBS and clinicians can be found in Appendix A.

Because conditions are rare, and interviewing providers on a series of multiple conditions is both time consuming and difficult given that not all conditions are seen in all places, it was decided to have interviewees relate the process of care to those disorders they see most frequently (see Appendix A for question wording). Interviews were conducted by a genetic counselor, and recorded. Interview notes were recorded on the interview tool by the interviewer. Summaries were then compiled. Recorded interview transcripts were then reviewed by the research team and coordinated with the interview notes with feedback from the interviewer. Qualitative thematic review was then used to synthesize the findings. This was done by first outlining the care process for the sample disorder across a one-year timeline and documenting any resource usage. These findings were chronologically summarized. A second review of the transcripts was then completed examining the thematic objectives of the instrument, which were: The function of the newborn screening activities in the state, issues related to the process of care, barriers to the care process and NBS expansion issues. The barriers theme was then coded upon analysis for three themes that emerged from the data which were family and parent issues, administrative issues, and education issues.

All interview responses were anonymous and findings reported here are de-identified using coded interviewee responses known only to the research team to promote candidness of response.

## **Summary of Findings:**

Here we summarize findings from the interviews. It should be noted, however, that many of these results, especially those relative to the care process, focus on four disorders, PKU, MCAD, Tyrosinemia I, and an unspecified Urea Cycle Disorder. This was a natural byproduct of the interview protocol used. Other responses relate to the provision of care for metabolic patients more generally. And while respondents were asked to focus on care during the first year of life, many extrapolated to issues beyond that timeframe, especially when discussing education, barriers and opportunities. These responses were left in the results as they provided context and a source of rich information.

The sections below represent a summary of the key findings from each of the focal areas of the interviews.

### **State-level Newborn Screening Protocols:**

- Overall this functions well, however what each NBS coordinator does, who they do it with, and the degree of coordination varies by state.
  - o Intensity of efforts also varies by condition
- There may be opportunities to synthesize the process, specifically relative to the need for enhanced education (themed below) and treatment protocols (if developed/available).

### **Care Process:**

#### *Initial visit*

- Care intensity during the initial visit is widely variable and condition dependent.. Tyrosinemia I and the Urea Cycle Disorder were highly intensive and time sensitive during the initial visit.
  - o Care processes can vary somewhat by disorder given intensity and urgency of follow-up
  - o Intensity is based upon the immediacy and location of follow-up after a positive screen.

#### *Secondary/Follow up visits*

- Resource use (i.e., visits, time spent on patient care, testing) varies dramatically after initial visit.
  - o Determinants are severity of the patient, knowledge of the family (discussed below), availability of providers, availability of dietary formula, and provider education.

#### *Laboratory work*

- There were no reported inconsistencies relative to lab work. The need for testing, when to test, and what to test for was consistent and agreed upon.

#### *Administrative and non in-person contact time spent on patient care*

- Administrative tasks were uniformly viewed as both substantial and complex.
- Issues cited were complexity of insurance protocols, the diversity of patients, parental and family education, and inadequate staffing.
  - o Staffing issues often created ambiguity as to who the primary contact person was, both from the perspective of the patient, but other providers. In some cases, the nutritionist served as the primary familial contact (and for certain disorders). At other times it was the geneticist.

- Those having the most frequent patient contact often ended up providing patient and family education and at times advocacy (for example with insurance issues).

## **Thematic Findings:**

### **Theme 1: Family/Parent Issues**

- Family and parental issues varied with the degree to which they had prior knowledge of or experience with a disorder.
- Education was a primary component. Depending on the condition, parents have to absorb a tremendous amount of information relative to treatment, monitoring, diet, insurance, care coordination and process.
  - Because some conditions can be so disruptive, parental response can at times be emotional and/or overstated, which was related as an important, albeit infrequent issue especially given the need for parents and families to often engage in the care process quickly.

### **Theme 2: System of Care Issues**

- Utilizing a team approach to care was considered a positive by all interviewees.
- Realizing a team approach was hindered by inadequate staffing.
  - Care teams differed substantially in their makeup depending on who was either practicing or available in a given area.
  - At least one provider indicated that inadequate staffing has an impact on providing high quality care.
- Tension exists between short staffing/lack of knowledge and not having enough patients with each condition to build knowledge/justify increased staff (discussed below in interview themes 2-3). For example:
  - In one state there were six babies in a year who were detected on NBS; 2 in another:
  - The resources needed for follow-up of false negative tests.
  - In Portland, Maine there were not enough patients to utilize the three physicians there.
  - There is currently no metabolic physician located in New Hampshire.
  - Staff adequacy seems to be caught between the intensity of care needed by these children, described above, and the number of children actually needing this intense care.
- The need for care protocols was discussed.

### **Theme 3: Education Issues**

- Education was reported to be one of the primary issues relative to care for metabolic patients.
- The need for enhanced education was mentioned with respect to families and parents, community service workers, and health care providers.
- Information provided families and parents was reported as often inconsistent and at times contradictory, causing confusion and anxiety on the part of the parent.
- Primary care provider involvement was viewed as inconsistent. The role of the PCP varied by patient and condition. Some viewed PCPs as engaged while others viewed the PCP as handing off patients to a separate care process.
- The need for additional supports to aid in education of metabolic patients and families was mentioned. Using genetic counselors was specifically mentioned.
- Examples of successful, although unfunded, educational solutions were mentioned, including a family educational camping weekend, and using schools as a point of coordination later in life.

- Respondents noted the need to coordinate locally, regionally, and nationally on care processes and best practices.
  - o Information dissemination was viewed as difficult due to the rarity of the disorders and their time spent treating them.

**Theme 4: Perceptions of Issues related to NBS Expansion**

- Providers were guarded to mixed about increased NBS expansion.
  - o Physicians felt doing so would add workload, exacerbate staffing issues, and augment existing other problems, especially for high resource intensity conditions.
  - o Mentioned were the 1999 expansions and increased workload that resulted.
- Nutritionists expressed concern that problems would be enhanced if the conditions added had a strong dietetic treatment component.
- All expressed ethical concerns
  - o Screening for conditions that have no treatment was mentioned, as was screening for conditions that have less pronounced effects on the patient. Both were seen as causing undue duress to parents and families, with the latter imposing more downstream resource implications relative to parental and family education.
- Despite these issues, many providers were optimistic about the ability to detect and treat additional genetic disorders.

Overall, interviewees expressed concerns that care for metabolic patients was widely variable depending on the condition, but relatively intensive upon detection. Issues post the initial treatment period revolved more around care coordination and education, both of the parent and family, but also of providers. Throughout the process, administrative time was viewed as burdensome and uncoordinated.

**Detailed Analysis Findings:**

The analysis below is divided into two parts; those related to the process of care from initial screen to the first year of life, and those related to themes drawn from the interviews more generally.

**Part I. Process of Care from Positive NBS to First Birthday**

Interviews revealed substantial variety in the process of care during the first year of life for newborns that screened positive for a metabolic condition on NBS. The processes described varied at all points, from the steps taken by NBS coordinators when they are initially contacted about a positive screen to the frequency of contact with clinicians and laboratory, and across multiple dimensions, including the type and severity of the condition and the characteristics of the patients’ families.

**I. NBS Program Role**

*Initial Positive Screening.* When the lab finds a positive or out-of-range result, they call the NBS coordinator (C0317, A049, A054, A038). Two NBS coordinators receive the probability that the result is a true positive (A049, A038) and 4 described receiving information about the condition, severity, and need for follow-up (C0317, A049, A054, A038). The type of condition detected impacts the pace at which this part of the process occurs (C0317, A038, A054, A049).

*Depending upon what the condition is that we’re trying to work...I contact the primary care physician and let them know what’s going on and suggest a course of action that takes into account the worst-case scenario. So, for example, if it’s possible that the baby has a fatty acid oxidation disorder, this child clearly can’t be allowed to fast or they may not survive the night,*

*that's how serious it can be. So I recommend that the primary care [provider] contact the family, explain what's going on and the baby not be allowed to fast, and that bloods or whatever it is, will be obtained to clarify the diagnosis. So we may implement some preventive emergency care measures until we know what's going on. (A038)*

*The lab usually calls me with what they report out as clinically significant NBS results. They usually provide me some guidance as to the level of urgency, you know they have a way of letting you know what the probability of a disorder is, which helps you to know how you're going to approach it in terms of urgency. (A054)*

*Based on those results and the recommendations from the screening laboratory, for example, if they tell us, the tests results, the likelihood of the condition, the impact on the health on that child and the recommended next steps for further evaluation. Whether it requires an urgent referral to a metabolic center or whether a non-urgent consult or just a repeat of the newborn filter paper is adequate. (A049)*

Two described repeating the test to confirm the positive screen (C0216, A038) while one stated that the positive result is confirmed at the metabolic clinic or with the primary care physician (A049).

*In general, the very first thing to do once there's a positive screen is to confirm that positive screen. One of the things that the New England Newborn Screening Program has done through the work of [referenced doctor] is to come up with probabilities based on the degree to which something is out of range. So I will get guidance when I'm called with an out of range thing that there's like a less than 10% probability that this is the real deal or there's a greater than 95% probability that this is truly whatever the condition is. That helps very much in terms of what comes next. In confirming the diagnosis, the next step might be to get a repeat filter paper and often times that's normal, there was some transient elevation for whatever reason and that's the end of it. Other times, depending upon the condition, how out of range, and what the family history is, that's a key component that we don't always know, the next step might be diagnostic testing such as sending plasma [testing] out to [the] clinical lab. (A038)*

In one state, if the confirmatory screen is also positive, the patient is sent to a contracted treatment center for all care and the positive finding is sent to a national database (C0216). However, in all of the other states, NBS coordinators are in touch with primary care physicians and clinical geneticists in order to coordinate care for the patient; two coordinators described being in contact with both (C0317, A054) while two others stated that she and the geneticist decide together who will call the primary care physician (A049, A038). Phone calls with clinical geneticists were used to discuss the result (C0317, A049, A054, A038) and the next steps in caring for the patient (C0317, A049, A038). Four NBS coordinators discussed faxing information, including information about the care plan and fact and ACT sheets, to the primary care physicians (C0317, A054, A038).

One NBS coordinator stated that at least sometimes she had direct contact with the family (C0317, A049) while another stated that they do not communicate with the family at all (C0216).

#### ***NBS Program Long-Term Follow-Up.***

All 5 NBS coordinators discussed NBS program long-term follow-up (LTFU). Three NBS coordinators stated that they had no in-house LTFU (C0317, C0216, A054) while a fourth state is in the process of implementing LTFU in-house (A049). One NBS coordinator stated that she received no information after the diagnosis was made (A054). One state contracted their long-term follow-up to centers within the state (C0216). Certain centers were contracted for the care of certain conditions; these contracts require centers to provide feedback on the patients detected on NBS. The fifth coordinator

stated that their program attempts to keep track throughout the diagnostic process and until the patient's 21<sup>st</sup> birthday (A038):

*I will keep tracking and keep on top of what's happening through the diagnostic process to see that diagnosis has been made. And...if it's a metabolic disorder, most often that child will be seen in the Metabolic Clinic...Right now, we don't have a nurse in the metabolic clinic so I fill both roles. So that [the clinical geneticist] and I will meet with the family initially together to do an assessment and explain the disorder, enroll the child in the metabolic clinic and that child will be followed on a regular basis, whatever that might be, by a nutritionist, a social worker, [the clinical geneticist] as the physician, me as the nurse, and we also have a psychologist who's available for that aspect. And, through that process, I get to know what's going on with the child not just through the first birthday but to the 21<sup>st</sup> birthday. It's a little bit different for kids who are followed in a clinic out of state and I don't have as much contact with the metabolic clinic folks. If they're assisting financially with the care, I may get regular reports from out of state clinics; that's sometimes problematic, but we would be able to find out what's going on with the child, with the parents' permission of course, by contacting that other clinic. (A038)*

## **II. Care from Clinical Practitioners**

Interviewees were asked to describe the continuum of care for a typical patient with a metabolic condition detected on NBS seen in their practice. Providers were asked to choose one metabolic condition in order to anchor their description of the process of care. Seven practitioners described the process of care for children diagnosed with PKU (5 clinical geneticists, 2 nutritionists), 2 described the process of care for a urea cycle disorder (1 nutritionist, 1 genetic counselor), and MCAD and Tyrosinemia were each described by 1 clinical geneticist. Two practitioners (1 psychologist and 1 nutritionist) did not specify conditions. Table 1 contains information on the processes of care described by clinicians.

### ***The Initial Visit.***

After a child has screened positive for a metabolic condition on NBS, the type and severity of the condition and parental anxiety impact the urgency of contact and treatment that a patient receives. Patients diagnosed with Tyrosinemia I, for example, have a concentrated but resource-intensive initial visit. As reported by one clinical geneticist:

*We bring the family in through the emergency room, get the child admitted, recruit all the specialists that's involved to evaluate the patient, which includes liver, you know every specialty, get our metabolic nutritionist on board. We work with the lab closely to confirm the diagnosis immediately, you know, get the result ASAP. And then comfort the family; initiate the treatment, which means we have to get prior authorization for the medication, talk to our pharmacy, talk to our nutritionist to get the food, to get the formula; teach the residents; get back to the primary care...And then after that couple of very intense days, the child is discharged. (A043)*

Similar intensity was described for patients diagnosed with a urea cycle disorder (nutritionist, genetic counselor) (B0414, C0317). One nutritionist described this first visit as "all consuming." Citing one specific case:

*I think it came back like the 5<sup>th</sup> day after birth, the NBS. The baby didn't have any symptoms yet and happened to be in the pediatrician's office, so we called over there and they came into the hospital. So from that point on, it's pretty intense...Basically, everything stops when I get a NBS like that, that's very nutrition labor intensive. My whole clinical, inpatient, outpatient, it's kinda on hold...[The initial visit is] all day. I'm kinda like, whenever there's not a doctor in there, I try to run back to see if I can talk to the family and get information. It's all consuming. It's all consuming. (B0414)*

Practitioners stated that patients diagnosed with a urea cycle disorders and/or Tyrosinemia I could be admitted to the hospital for anywhere from a couple of days (A043) to up to two weeks (B0414) and their care involved many contact hours by providers. The clinical geneticist who described Tyrosinemia I stated that she was involved with the patient 3-4 hours on the first day of the admission (A043) while the nutritionist stated that the urea cycle patient required 6-8 hours a day each day of the patient's 2-week admission (B0515).

**Table 1. Process of Care Described the Interviewees:**

ID	Role	Condition Identified	Visit Length and Frequency				Laboratory Tests
			Visit 1	0-3 months	3-6 months	6-12 months	
A027	Clinical Geneticist	MCAD	1-1.5 hours	visit 2: 30-45 minutes; then varies; usually follow up at 1 month	variable; usually follow up at 3 months and 6 months	variable	visit 1: bloods; 6 and 12 months: carnitine; some have DNA analysis; sick labs
A0510	Clinical Geneticist	PKU	1 hour (md time)	visit 2: 1 hour (md time); revisit 1 week, then 2-3 weeks, then monthly: 45 minutes (md time)	every 2 months: 45 minutes (md time)	every 3 months: 30-40 minutes (md time)	visit 1: plasma amino acid analysis (short phenylalanine and tyrosine and overnight full amino acid analysis), filter paper, filter paper urine; visits 2 and 3: blood phenylalanine level rapid analysis; parents do weekly filter papers (sent to lab); labs done at all visits from visit 3-12 months old
A021	Clinical Geneticist	PKU	4 hours	weekly for 4-8 weeks, then every other week (not seen by clinical geneticist)	every month	every other month	visit 1: amino acid, urine, blood; parents weekly blood draws (sent in)
C013	Clinical Geneticist	PKU	1 hour (md time)	seen by whole team as soon as it can be arranged: 1.5 hours; 1 additional visit with clinical geneticist within 6 months; 3-4 visits within first year: 45 minutes - 1 hour			diagnosis- urine and blood; filter papers: weekly, biweekly, monthly sent from home; some have neuro-psych and/or genetic testing
A032	Clinical Geneticist	PKU	2-3 hours	monthly, 1-1.5 hours	monthly: 1-1.5 hours	every month or every other month: 1-1.5 hours	blood tests twice a week; Co-factor screening: 1-2 months old
A016	Clinical Geneticist	PKU	1-2 hours (md time)	Monthly	monthly		Repeat NBS screening; weekly filter papers: at center until about 6 weeks, then done at home
B0515	Nutritionist	PKU	4-6 hours total (2 hours nutritionist time)	visit 2: 30 minutes-1 hour (nutritionist time); weekly to 6 weeks, then monthly: 30 minutes (nutritionist time)	monthly: 30 minutes (nutritionist time)	every other month: 30 minutes (nutritionist time)	confirmatory testing; weekly bloods (sent in); amino acids testing done periodically
B0212	Nutritionist	PKU	2 hours	monthly clinic, home visits between clinic visits: 1.5 hours	monthly clinic visits; home visits between clinic visits	varies	Repeat NBS screen; blood tests: weekly blood test for 3 months, then bimonthly blood tests; more frequent blood work when child is sick
A043	Clinical Geneticist	Tyrosinemia I	ER admission (3-4 hours md time)	weekly for the first month or 2, then bimonthly visits, then monthly by 6 months: 45 minutes-1 hour			monthly, then every couple of months blood and urine at each visit
B0414	Nutritionist	Urea Cycle Disorder	hospital admission (6-8 hours nutritionist time each day for 2 weeks)	visits 2 and 3: within the first week; then weekly or bimonthly: 30 minutes-1 hour	weekly or bimonthly, 30 minutes-1 hour		plasma amino acids, ammonia via blood
C0317	Genetic Counselor	Urea Cycle Disorder	hospital admission (3-4 hours gc time)	weekly, bimonthly, monthly: 1 hour			molecular testing
B0111	Nutritionist	no condition named	2-3 hours (formula treated patient)	depends on diagnosis: about every 2-3 months: 30 minutes (MD), 30 minutes-1 hour (nutritionist)			confirmatory testing, fibroblasts, skin biopsy
B0313	Psychologist	no condition named			visit at 6 months: 1-1.5 hour visit	visit at 12 months: 1-1.5 hour visit	does not order labs

Patients diagnosed with MCAD and PKU were also seen quickly, although they were not admitted to the hospital. Here two of the four clinical geneticists describe the process of seeing patients with PKU within a few days of the initial positive screen (A0510, A027, A021, A032):

*It's usually the nurse or nurse coordinator who takes that call [call from pediatrician or NBS laboratory] and sets up the appointment, which is usually done within 24 hours. ...if we get a call on Friday, it's generally for the next appointment day. If I get called or if it's an inconvenient time, then I will speak to the pediatrician directly. If it's for a Friday, [and], if in their opinion the patient or the family is going to be very nervous and anxious, I'll usually call them that afternoon or just to give them a heads up on what I'm going to be doing and the fact that the child is in no danger. We'll have a, sort of a, counseling session on the phone. But if the child is to be seen within about 24 hours and I'm called by the pediatrician, usually...we just make sure the appointment is set up. (A021)*

*Depending on what that initial level was, like if the initial level was 7 or 8 or something, we're going to do things a little bit differently than if it was like 4. So, if we're assuming that it's high on the initial screen, like a 7 or an 8, usually I'll have the primary care doc[tor] see that family that day or the next day, get a repeat [screen] card. They'll talk to the family a little bit about what's going on and we'll see the family within a couple of days, usually before that repeat card comes back. If the level started off at like 4 or something, we kind of do the same thing, except we may wait a little bit longer...before we see [the child] in clinic. (A032)*

When screened levels were lower and less concerning, two clinical geneticists (A027, C013) described seeing patients who screened positive for MCAD and PKU, respectively, within the week of the phone call from NBS, not within a couple of days:

*...If I'm more concerned [about likelihood and severity] I will see the patients, sometimes that day or the next day. Or if it's looking more to be something like a carrier or a false positive, I will see them in my regular clinic, which could be the next day or the next week. (A027-MCAD)*

*If the initial is high enough, she [the state's NBS coordinator] also calls all the other members of the team...the social worker and the nutritionist. Usually one or both of them visits the child in the hospital or in the home, depending on where in [in the state] the child is and do a home visit to get formula to them. I see them probably in the first, well, within the first week. (C013-PKU)*

Initial visits for patients with PKU and MCAD were shorter than initial visits for Tyrosinemia I and the urea cycle disorder. The initial visit for MCAD was reported to be approximately 1 to 1.5 hours long, not including lab work or meetings with other members of the health care team (A027). The length of the first visit for PKU varied; clinical geneticists reported that the initial visit lasted from 1 hour (C013, A0510) up to 4 hours (A021); nutritionists described the first visit of patients with PKU lasting between 2 (B0212) and 6 hours (B0515). Providers also described laboratory tests collected during this first visit (A0510, A043, A021, C013, A016, B0515) and 6 clinical geneticists discussed introducing the patient to other team members (A0510, A043, A021, C013, A032, A016). Two clinical geneticists and one nutritionist also described trying to make these visits more efficient by sending patients for lab work at the beginning of the visit so that it could be processed during visits with the clinical geneticist and ready for the visit with the nutritionist, which added to visit duration, but also addresses the need to obtain information quickly in some cases (A0510, A021, B0515):

*We will see the baby again in 3 days, at which time we'll immediately measure the blood phenylalanine level. Again, have the family, before they come up to the floor...go to the laboratory and get a blood specimen from the baby, and have that taken over to the lab.*

*Sometimes I'll go over to the lab myself and take the blood specimen by hand up to the laboratory. I will have called the laboratory ahead of time and warned them that we're going to have a blood specimen delivered which we want to be analyzed for phenylalanine and tyrosine as quickly as possible which means that they'll set up themselves before I get there, putting the control line and so forth. So by the time I get there, they're ready to put the specimen on and we can get that blood phenylalanine level back within maybe an hour and a half. Meanwhile the family will go back up to the clinic. (A0510)*

### **Subsequent Visits.**

Patients diagnosed with a metabolic condition on NBS are seen frequently during the first year of life, although the frequency and length of these visits varies by condition, severity, and family characteristics. The second visit often occurs within one week of the initial visit (A0510, A027, A043, A021, A032, B0515, B0212, C0317). Patients with these conditions were reported to then graduate to less frequent visits (A027, A0510, A021, A032, B0515, A043, B0414, C0317). For example, one clinical geneticist described how PKU patients were initially seen weekly, then every other week, and then each month before finally being seen every other month at six months old (A021). This graduated process was also described by practitioners discussing both the initially most urgent conditions (the urea cycle disorder and Tyrosinemia I) and the initially less urgent conditions (PKU). Follow-up visits lasted from 30 minutes long (A027) to at most 1.5 hours long (B0111, C013, A032, B0212).

Some care processes were more difficult to describe than others. Practitioners spoke of the fact that it was difficult to describe the process of care because it depended upon factors related to the family. Three clinicians described the impact that family anxiety, understanding of the disorder and/or prior experience with it, and the need for support had on the frequency of contact (A027, A032, B0212).

*...I usually see them back within 1 week to discuss the results of the studies that were done at that previous visit [visit 2]. Then it all depends what those studies showed and occasionally, for MCAD, I usually either know or I don't know but there are those panic, grayer cases where I have ended up sending DNA analysis, for [a] mutation. Then usually, again depending on severity, if they're more severe or I'm more concerned, I might see them back in another couple weeks, making sure that they're doing the protocol that they're supposed to be doing [and] the baby's growing well. It's really hard because, from there, it's just really individual cases. It's a case by case basis. If I feel the parents get it, then they might get an extra week; if I feel that the parents are a little unsure and they're nervous and they're scared, then they come back and see me the next week. It's just parental anxiety....I usually see them in another month, and then when they're about 3 months, and then they're six months. If they're growing well and they're doing well and the parents are doing well, I just am periodically checking with them every few months through the first year. (A027)*

*A lot of it is sort of how much support the family needs. If it's their second kid with PKU, they may not need the same amount of support. If it's their fourth kid, the first one with PKU, they may not need the same support. If it's a young family, first baby, mom's 18 kind of thing, they need more support. So we...gauge it. (A032)*

*After that [first six months of treatment for PKU] it varies... I do home visiting between times, and that's not on any kind of a regular schedule. A lot of it depends on how the family's doing and how close they are...A lot of it depends on...how the family is able to handle this diet because if the family is pretty together and understands things, we don't need to be on top of them every minute. (B0212)*

Any patients who screen positive for metabolic conditions on NBS are also often assessed for developmental delays; this assessment was described as including 2 visits, at 6 months and 12 months of age (B0313).

#### ***Laboratory Work.***

All interviewees except the psychologist described the need to order laboratory tests. Common tests included urine tests (A021, A0510, C013, A043) and blood tests (A021, A032, B0515, B0212, A0510, C013, A043, B0414, A027). Practitioners noted that many of these tests were done weekly during the first year of life. Families of children with PKU were often instructed to have lab work conducted weekly and to send the collected specimens to the laboratory for testing (A0510, A021, C013, A016, B0515, B0212).

### **III. *Administrative and Non-In-Person Contact Time Spent on the Care of Patients.***

Time spent on administrative tasks relative to patient care was reported to be substantial. These tasks included time spent on the phone with the patient's family and pediatrician, doing case management and dealing with insurance companies (see Table 2). One clinical geneticist stated that she could not count the number of hours spent doing these tasks (A043):

*Hours. A lot. I think it's a little difficult for us to quantify. We have nothing to quantify it with systematically, but I can say 'a lot'. (A043)*

Two clinical geneticists stated that the amount of time spent doing these tasks was high during the initial period of diagnosis and then tended to taper off (A027, A032). Clinical geneticists estimated that they spent several hours on the telephone with the family and pediatrician per patient who screened positive for a metabolic condition in the first year of life; one clinical geneticist reported spending 3 hours dealing with insurance companies and another 5 hours on administrative tasks in the first year (A0510). A total of five hours spent on administrative tasks in a year was low compared to the reports of other clinical geneticists who reported spending one hour per visit (A021) and one hour per week respectively (A016).

Nutritionists spent time on administrative tasks, but primarily through phone contact for patients treated with dietary intervention. Nutritionists had frequent and, sometimes, immediate contact with families (B0414, A032, B0111):

*When the patient does go home, they usually go home on a Friday or something. The parents are usually in contact with me on the weekend; they're usually in contact with me any time for the first 2-3 weeks. I give them my pager and my cell phone and they call me anytime. (B0414)*

*The nutritionist and clinic coordinator will usually follow up with them [the family] the next day [after the first visit]...daily for a couple of days to see how things are going. Usually, have them back in the office in a week or do a home visit in a week. (A032)*

*To some degree...I serve also as one of the primary contact persons, staff people in our clinic for outpatient questions or concerns. So that's another kind of role that I take on...depending on ...the disorder. If it's PKU, then certainly I'm talking to them pretty often, sometimes it's weekly with levels and diet changes. (B0111)*

Nutritionists reported spending at least 1 hour per week on the telephone with families and pediatricians (B0515, B0111) and at least one hour per month on administrative tasks (B0515). One nutritionist reported spending less time on the telephone with the family and pediatrician but reported spending 20 minutes spent per blood test administratively (B0212). Considering that these blood tests are repeated weekly for 3 months and then every other week after 3 months, the reported 20 minutes equates to

roughly 360 minutes or 60 hours of administrative work per case during the first year of life (B0212). As one nutritionist described, the work beyond the visit is time consuming (B0414):

*The visits not as long as the work I have to do to make the changes. So [during the visit] they come for half an hour to an hour to our clinic...I'm...still putting in at least 2 hours afterwards. Two to three hours to recalculate the formula, well actually to do the intake, to just make the changes, contact the family, email the prescription, write up the note, order the formula. (B0414)*

**Table2. Time Spent by Clinicians Per Patients Doing Work Outside of Visits**

ID	Role	Condition	Time Spent				
			On the Phone with Family Between Clinic Visits	On the Phone with Pediatrician Between Clinic Visits	Doing Case Management	Dealing with Insurance Companies and Other Admin Tasks	Administrative Tasks
A027	Clinical Geneticist	MCAD	"a couple of hours" [in the first few months]			varies	couple of hours per year
A0510	Clinical Geneticist	PKU	1 hour between visits	15 minutes total	3 hours total	3 hours total	5 hours total
A021	Clinical Geneticist	PKU	(no answer)		none	none	1 hour per visit (geneticist); 1-2 hours per visit/blood level (nutritionist)
C013	Clinical Geneticist	PKU	couple hours per year		(no amount of time given; often done by team)		1 hour per year ( 4 hours per year: VLCAD)
A032	Clinical Geneticist	PKU	couple of hours per week for the first couple of weeks and then it slows down substantially				
A016	Clinical Geneticist	PKU	several hours initially; rest of the time: 40% of total time				about 1 hour per week
B0515	Nutritionist	PKU	1 hour per week		(no answer)	3 hours initially, then less	1 hour per month
B0212	Nutritionist	PKU	30 hours per year		monthly meeting	5 hours per year	20 minutes per blood result
A043	Clinical Geneticist	Tyrosinemia I	could not quantify the amount of time spent				
B0414	Nutritionist	Urea Cycle Disorder	2 hours per month		2-3 hours per week	sometimes	30 minutes per week
C0317	Genetic Counselor	Urea Cycle Disorder	2 hours over 10 months	(no answer)	(no answer)	(no answer)	more than 5 hours over 10 months
B0111	Nutritionist	no condition named	at least 1 hour per week	none	a lot	1-2 times per year	infrequently
B0313	Psychologist	no condition named	(no answer)	(no answer)	(no answer)	2 hours per year	1.5 hours per visit

## Part II. Interview Themes

As one clinical geneticist stated there are “barriers at all levels” (A021).

The following are four themes that were referenced across interview respondents. They are Family and Parent Issues, System of Care Issues, Education Issues, and Perceptions of Issues Related to NBS Expansion.

### Theme 1: Family/Parent Issues

Health care professionals spoke about a myriad of issues they had dealing with the parents and families of children who screen positive for a metabolic condition on NBS. Once a diagnosis is made, parents often lack knowledge about the condition (B0212), are anxious about the unknown (A043), and have questions regarding the costs of diet (A0510). Providers further face problems with compliance, including parents’ difficulty managing the diet (B0212). Two providers described resistance to follow-up care (B0313, C0317); as one provider stated (B0313):

*With the MCAD, they’ve been so traumatized by the process of identification, they don’t want to come back to the clinic. We have a couple there, that say, “I come and you don’t do anything”, especially the ones that are just, you know, don’t know the facts, that just take them in if they get sick. They go “I come to clinic, what do I get from clinic?” So they don’t want to come to clinic. They’re not getting a treatment, they get the baby stuck. “Why should I get my baby stuck?” (B0313)*

One geneticist described the issues she faces with parental compliance in the following way, but explained that these parents are the exception rather than the rule (A027):

*We have some parents, for example [with respect to] PKU who are so incredibly type A, that we over-restrict...they’ve marked everything little thing down and they keep track of every single level. And then I have had another child that I’ve had to admit to the hospital twice to see why I can’t get her levels under control because I thought it was home environment. So, parent compliance and understanding [is] hard sometimes. (A027)*

Certain parental characteristics also made caring for children with a metabolic condition detected on NBS more difficult. Interviewees mentioned that language barriers make providing health care to these children more difficult (A043, B0414, C0317); for example, one nutritionist stated that language barriers specifically “make the process three times as long at least, and it’s never really as good” (B0414). Financial barriers, including the costs of frequent visits were also stated as a problem (A043, B0212, B0515). Families that lacked social supports (B0212), or were teen moms (A027) or single parents were also more difficult to work with (A043). Because of the location of providers, geography can also be a barrier for families who need care but live far from providers (A043, A032, B0212, B0313, A027).

### Theme 2: System of Care Issues

Interviewees also spoke about many of the issues within the system of health care for children who screen positive for a metabolic condition on NBS. Caring for patients using a team approach was mentioned by all professionals; however, the team approach is less likely to occur due to low staffing and a lack of system-wide synergy. Practitioners also mentioned day-to-day difficulties that made the system less effective, including problems they encountered when dealing with insurance companies.

### *Team Approach with Inadequate Staffing*

Every interviewee (except psychologist) mentioned being part of a team that cared for children who screened positive for a metabolic condition on NBS. All interviewees mentioned the geneticist as a team member (100%, n=17); nutritionists were mentioned by 65% (n=11) and nurses were also mentioned by 41% of respondents (n=7). Fewer respondents mentioned social workers (24%, n=4), genetic counselors (24%, n=4), and parent advocates (12%, n=2). Psychologists, nurse practitioners, physicians, and NBS coordinators were mentioned by one respondent each.

Problems of staffing were noted to be both immediate and long-term. For example, one clinical geneticist mentioned that her clinic was without a genetic counselor for 3 years (A043) and others mentioned that they specifically lacked nurses (6%, n=1), social workers (12%, n=2), genetic counselors (18%, n=3), and nutritionists (6%, n=1). In general, respondents mentioned a shortage of personnel, including clinical geneticists, nutritionist, nurses, and genetic counselors. These staffing shortages have resulted in program cuts (B0111) and challenged providers in the field:

*I think here in [the city], we do fairly well and I think a lot of it is because we have a couple different people with very different expertise that can really help with things. [The other clinic site is] more difficult because I'm only there 2 days a month. I don't have access to the charts in between times because the charts are kept in [the other clinic] and I'm two and a half hours away. And, they let go a lot of the staff that was helping; we had a nurse that was very good, would take care of prior authorizations, call in prescriptions and all that kind of stuff. Now, I get all those phone calls and I have to call somebody to get them to fax the information from the chart to me so I can review it and figure out...so it just takes like five times as long to do anything because there's all these other steps that have to happen. We don't have the same kind of outreach home visiting kind of capabilities [there]. [The nutritionist's] time is much more restricted because she's only very part-time to do metabolic stuff. She's very good, and she's very good about weaseling it into her day and very responsive to families on the phone but it's just a different model. They're sort of a no news is good news...we call out all of the lab results on our kids, they don't. It's a time and personnel issue. (A032)*

*Now, if I were more than one person or we had a larger clinic, we probably would see them more often. (C013)*

As this clinical geneticist described, the need for adequate staff negatively impacts patient care. This clinical geneticist also presents another problem inherent in the system: determining adequate staff size. Staff adequacy seems to be caught between the intensity of care needed by these children, described above, and the number of children actually needing this intense care. Two practitioners spoke of the low number of children who actually receive a positive diagnosis for a metabolic condition on NBS (A016, B0212). A nutritionist estimated that there are only about six newborns detected with one of these metabolic conditions on NBS each year in her state (B0212); for every one case of PKU, there are two cases with other conditions detected (B0212). For example, Maine currently has only two clinical geneticists, a reduction from three, because they do not have enough patients for three providers (A016).

### *Need for System-wide Synergy*

Because the care for children with metabolic conditions requires a team approach, it is important that the system work synergistically. For example, one geneticist mentioned that transitioning patients to adult care when they outgrow pediatrics is difficult because there is no one to transition them to (A021). Another mentioned that at times, pharmacies do not carry the prescribed formula and a pharmacy that carries it must be located or the formula must be ordered (A032). One important aspect of system synergy is that the responsibilities of team members are delineated, especially as medical home is implemented (B0313), and that each provider works within his or her set of responsibilities. That this did

not seem to be the case was especially evident for nutritionists (B0414, B0515, B0111). One nutritionist described her role in following patients when they are admitted to the hospital this way (B0414):

*My other challenge is that when a patient comes in to [the hospital], one of my patients maybe comes in because they're ill or for some reason they're admitted. I pretty much have to follow them. So, again, it's not like there's a nutritionist who I hand off to who's inpatient. It's like I get the call, "...so-and-so's here", and it's me and then everything else gets dropped so I get that settled or figure out what's going on or make the formula, like I did the other night on my way out the door... There's no handoff [at the hospital]... it's just me. They would have in-house nutritionists but it's always been hands-off. And it's not that I mind even doing the management part, but it's very difficult to run back and forth to [the hospital] all day long. I'm not in the same building or the same place and that's difficult. And also, there's no person on-call for our formula room... its done shut down...but these babies can't wait (un)til the next day to have their formula. They'll die. They don't understand that if the order's not in by 3, they're just gonna have to wait (un)til the next day...you just can't do that. Somebody has to know how to do that that's here all the time. (B0414)*

This nutritionist is also responsible for ordering, inventorying and stocking all of the foods and formulas provided by the state, which takes an additional 20 hours per week (B0414). Another nutritionist describes working as an advocate and case manager for her patients. Especially with PKU, she (B0515) describes that knowledge is poor, and that parents cannot advocate for themselves to insurance companies. In general, she describes the confusion surrounding her responsibilities for the patient's care:

*Somebody thought that what we do is basically case management, you know, sort of being the triage person and working through a lot of these administrative issues. I don't think it's a particularly good use of a nutritionist's time, to be doing the insurance part of these things. We've learned over time to do it, but it's not the best set up. I think it'd be great if there were [some] sort of care management at some place, either within the pediatrics office or at the hospital to have somebody that really does more of that care coordination. We do it, but it's not really the best use of our time... We do have a nurse practitioner and she does a lot of this, we sort of share the wealth on this, but some parts of it require a nurse practitioner and some really require that somebody keep all the pieces together; it's almost like we need a coordinator at the clinic that is somewhat a health care provider but really a coordinator of all these systems. (B0515)*

Another system-wide problem discussed by health care providers was health insurance. Providers described this as the primary problem they faced (B0313, A032). Insurance issues cited were the complexity of insurance, that each patient's situation differed, and that insurance companies often change the coding of formula benefits (B0515, A032). Respondents noted that they spent a lot of time on the phone and writing letters of necessity given each insurance company's unique approval process (A027, B0515). One specific problem with insurance for children with metabolic conditions detected on NBS related to the costs of diet treatments covered (A0510). This problem varied across states, since states vary in their support for diet. For example, respondents reported that New Hampshire residents have limited financial support for diet needs (A0510) while Rhode Island's NBS program provides formula (A043).

Interviewees also discussed care coordination and information seeking issues within the system of care. Two clinical geneticists and one genetic counselor mentioned the need for more time, first, to find information on resources that parents might need (C0317), second, to give patients all the resources and information they need and want (A0510); and third, to meet all of the needs and coordinate care and services required for proper treatment of these children (A021). Funding was also a problem. Interviewees described needing money for resources (A032) and clinic personnel (B0111). They also

discussed a more fundamental problem; that the care of children with metabolic conditions is poorly reimbursed by insurance companies and hospital funding or other funding is often necessary to supplement the costs of care (A021, A043).

### **Theme 3: Education Issues**

Interviewees were asked specifically about education relative to metabolic conditions broadly defined. Four providers indicated that the current state of knowledge and awareness for metabolic conditions is poor (B0515, A021, C013, B0313) and one nutritionist stated that education can always be improved. However, two clinical geneticists believed that, although still poor, the receptiveness of primary care physicians accepting these conditions has improved over the past decade (A032, A016). Providers offered insight on (1) who they felt needs to be educated about metabolic conditions, (2) ways to enhance education, and (3) the impact of knowledge on their own practice.

#### *Who Needs to be Educated*

Providers highlighted two non-family groups of people who they believed needed to be educated about metabolic conditions: community services and health care workers. One clinical geneticist described the need to educate community service agencies and workers, especially as children grow older (A021):

*We also are involved in training community services, whether it's physical, occupational, [or] speech therapy, [as to] what they have to do [for these children]. We have to educate the schools; we have to educate the workplace...college campuses because often these patients need support from dietary services and educational centers. In the work place, sometimes there are special needs. In the schools sometimes there are non-dietary special needs that are required as well that they utilize the nurse. If the family is not an organized family, then often those support services—nursing supports, VNA support—are more intensive at times and we take on that role. Barriers... are lack of awareness and the amount of time that's required to do this and do it well. If you put the time in up front, it usually saves time in the long run. (A021)*

Two providers (A021, C013) described the need to educate physicians. One related the need for education to the ability of the primary care physician involved with patients who screen positive for a metabolic condition on NBS (A021):

*Community physicians are not well trained and so the role of the clinic is not only to educate the parents but also to educate their clinicians, which is time consuming. Sometimes it's successful, sometimes it's not. The primary care physician is often isolated as the patients or families learn how much easier it is to speak directly with the clinic and unless the clinic makes an effort or the patient's family makes an effort to keep the pediatrician involved, the pediatrician becomes sidelined and some don't react well to that. Some are very happy with that. And sometimes it involves disruption of that relationship where if the pediatrician can't be involved they don't want to stay involved or they feel that even if they're kept involved, they don't feel comfortable playing a role and so the patients have to end up looking for a new pediatrician, which basically the main criteria is someone who's not afraid to deal with complex patients and who feels comfortable picking up the phone and asking questions if they're not sure of something. (A021)*

#### *Ways to Enhance Education for Families and Communities*

A number of ways to improve education about metabolic conditions detected on NBS screening for families of patients and their communities were proposed by interviewees. One NBS coordinator believed that education could be enhanced prior to birth (C0317):

*One thing I've noticed repeatedly is that [there] is an increased educational component from the prenatal side of things, so whether that's the newborn nursery staff, prenatal nurses, OB/GYNs, or all of the above. I've had multiple families after a positive diagnosis say to me things like 'well our birth instructor mentioned it but said we shouldn't even focus on NBS because nobody gets a positive result' or 'oh the nurse came in to prick his heel but said you can forget I'm ever here, you know, this is a PKU test'. And I had a family with a urea cycle disorder who had significant difficulty acclimating to the fact that their child had a urea cycle disorder because they were convinced the test only tested for PKU. So I think there's just a learning challenge in the community that's bigger than the metabolism clinic. I've noticed it repeatedly in families who get very stuck in how it was presented to them and I think it would be a good area to help with the educational component. Even if they heard it correctly the first time and they don't remember it, it might improve their reaction to NBS a little bit... and it's not the same hospital or the same nurse; it seems to be kind of spread out (C0317).*

Two clinical geneticists stated that education in their clinics could be improved by the presence of a genetic counselor (A0510, A043). A clinical geneticist discussed providing families with resource binders containing contact information and when contact should be made; however, these binders need updating (A032). Consistent with their comments on family characteristics that make providing care for patients, one provider discussed the need for materials in different languages (B0414). The use of the internet by families was also discussed; the psychologist mentioned the need for more information on false-positives so that families do not turn to the internet as often, and two other providers mentioned the need for up-to-date, family-friendly and complete websites (A043, C0317). A nutritionist also discussed that families affected by metabolic conditions needed to be connected with other families through group sessions (B0515).

However, education improvements that have occurred were also discussed by interviewees. A clinical geneticist spoke of being part of another clinical geneticist's education grant; the grant funds in-school education for school nurses and teachers about the needs of students with metabolic conditions (C013) and another spoke of working closely with schools to make sure that dietary needs are met (A032). Another clinical geneticist spoke of a camp for families of children with PKU and allied disorders (A032):

*The other thing we do is we have camp every fall. We do PKU and allied disorders, so it's a low protein camp. It's a family camping weekend. It winds up being funded through fundraising and then we get support from a bunch of the food companies and formula companies. Families bring their own formulas but all the low protein food is provided. They stay in cabins and there's sort of educational things for the families, educational/fun things for the kids. It's \$50 for a whole family for the weekend, everything included. It's a lot of work-a lot of work-but the families love it (A032).*

#### *Information Needs: More Information, a Place to Get it, and Time/Space to Learn it*

Interviewees spoke of their own information needs. Specifically, they spoke of the need for more studies, standardized care guidelines, and the time to learn about the conditions. However, these information needs must be understood within the context of disease prevalence. In one state, only six children were detected with a condition on NBS (B0212- verify with umass #s). There are very few patients from which to gather knowledge in any given year, which limits the amount of information that can be gathered and the amount of experience that providers can individually gain from working with these children. One nutritionist described the problem she's having working with a team to develop evidence-based dietary guidelines; the problem, she stated, is that "there's not a lot of evidence" (B0515).

Health care providers spoke of the need for studies to understand the conditions and some ongoing projects addressing information gaps. Individual providers spoke of the importance of studies on

genotypes (B0313) and gathering information on the severity (B0515) and developmental delays (B0313) associated with metabolic conditions detected on NBS. Providers also spoke of on-going projects to improve knowledge, including a nutrition guidelines project (B0111, B0515), a project on delays in parental bonding (B0313), and a project on analyzing care (C013). One NBS coordinator also posited the following:

*I don't know how many states have board certified metabolic physicians. And I don't know how helpful it would be to physicians who are practicing as consultants without being board certified...to offer additional training to them without having them go through the certification process, which is lengthy and expensive. In a place like New England, where our populations are small, it may not be cost effective to go through certification because you just wouldn't see that many patients in a year. So I think making available specialized metabolic training for the physicians and the clinical follow up staff in metabolics would be great. Something on a local level that doesn't require flying off to Baylor in Texas or Wisconsin. My other concern, in general, has always been what happens to a patient with a metabolic disorder after they hit 21. At least in our state, [the state program for children's health] offers the only metabolic clinic services in the entire state and because we're funded by Title 5, [their coverage ends] at age 21 they're no longer children, with some limited exceptions for women of childbearing age. And I've always been concerned that there's no low-cost or free adult metabolic clinic or support services, like for an adult male with PKU who's 22 years old. You know, what happens? They're reliant upon whoever their primary caregiver is and they might not get accurate information or follow-up. (A038)*

One nutritionist stated that getting answers to these questions and sharing information either nationally or regionally is essential (B0515).

Three providers spoke about the need for standards of care. One clinical geneticist stated that there needed to be consensus on treatment (C013). Nutritionists especially felt that they were at a disadvantage because of the rarity of these diagnoses and the need for standards of care (B0414, B0212). Two nutritionists stated that they have not worked with all of the metabolic conditions. They described their on-the-job learning process and what would be useful to them:

*The thing that's interesting about the field is that I might be considered the specialist here, but I haven't seen that many. I mean every time you get a NBS that's something really different, you've never seen it before and you have to start from the beginning learning it...It's like okay, I've done MSUD and PKU, but every time I get one variation propionic academia, glutaric academia, whatever the diagnosis is, you're thrown right in. Unfortunately for me, I don't really have any other nutritionists that I work with...I have some parent liaisons here at the clinic who are very helpful in terms...if there are insurance issues. We also run a support group once a month for PKU patients and they helped me start that. But really every time you get a new diagnosis...[there is] anxiety and bringing the book home with me on the weekend, and [someone from the clinic] calling me at home on the weekend. So once someone serious comes in, [someone from the clinic] will call me and say 'okay I think you should start reading about this over the weekend'. It's just at the drop of a hat and I'm afraid to take a vacation. When things are slow it's great, but then all the sudden it's like boom. (B0414)*

*Our protocols are kind of individual. One of it's a function of the fact that we just don't have that many kids. All of us are skilled practitioners in working with kids with chronic health conditions so we all know and are able to figure out quickly what families need. We know metabolic disorders enough to work with it. But we don't see enough kids that we, at least I, feel as though I need some advanced protocols and what I need to do. ...It would be really nice to have*

*some...protocols out there, but...as I said, there's a lot of kids I've never seen. So if a kid with maple syrup urine disease gets diagnosed in our state, I'd be panicking, trying to figure out, alright, where do I look at this, where do I look at that. So, I either talk to [other nutritionists in the region] and they guide me through... (B0212)*

#### **Theme #4: Perceptions of Issues Related to NBS Expansion**

Interviewees were also asked how they thought that the further expansion of NBS would affect their practice. Many of the concerns expressed reflected current issues that providers were already experiencing: low staffing, added workload, constrained time, and concerns about education. Expansion also met with ethical concerns about the treatment and utility of adding new conditions.

##### *Impact on Current Practice*

NBS expansion concerned geneticists differently than nutritionists. Although one geneticist stated that the impact on her practice would depend on what was added (A027), in general, clinical geneticists thought that expansion of NBS would substantially impact their practices by making them busier (A032, A0510, A043) and increasing work load (A032, A0510).

*Very significantly. If for instance, if we get into the lysosomal disorders screening and other kinds of screening, we're likely to see the children who are identified in the NBS with the possibility of having a lysosomal disorder. And that's going to mean that we're going to have a lot of questions in our own practice with regards to how do we follow-up these families, whether they need treatment or not, whether we should refer them to somebody who has more experience in lysosome disorders or whether we're going to get into that experience with an infant who has been identified as a possible lysosome disorder on NBS. And discussion with the families is going to be added time to our clinic practice...It's definitely going to add very significantly to the time of our clinic practice. (A0510)*

*It's only gonna get worse. It's...very time intensive and they're unpredictable. And that's the problem. If we keep adding to [the number of] conditions, there's going to be more emergency follow-up... and that's going to get more stressful. Especially when there's not necessarily [anything] we can do. (A032)*

One clinical geneticist cited the difficulty of incorporating care for patients with conditions added to NBS in the 1999 expansion in Massachusetts as a model for further expansion (A021):

*When NBS was expanded in Massachusetts in 1999, it was assumed that the practices would simply absorb these patients. You know, we are preparing for the next round of expansion which includes the lysosomal storage diseases, and I think on some level it's great for the patient, it's going to be a disaster for the clinic. Because... that one identified patient, given how quickly things have to move and how intensive the work is that has to occur, that can take up your time for the week. You just have to put everything else aside. Given the...lack of financial supports, given the climate of the fact that we don't have enough clinicians...I think we're already seeing patients leaving or clinicians leaving health care to work in [other] industry or other areas because they say it's too hard and I think it's going to aggravate that. Ultimately, if that happens, of course, it's not going to bode well for the patient in the short-term or the long-term and it's not going to bode well for the profession. (A021)*

Nutritionists were less sure about how NBS expansion would impact their practice; one stated that she had “no idea” (B0212) and another stated that the impact would differ based on the conditions added and whether or not those conditions would be treated with dietary regulation (B0515). However, one nutritionist felt that expansion would be more labor intensive (B0414). Three providers (a geneticist, psychologist, and nutritionist) stated that expansion meant the need for more staff (C013, B0313, B0111):

*I think the infrastructure thing is a big one if you're thinking of NBS. Because it's wonderful; it seems like the expansions are just booming for a lot of programs around the country, in the US and probably internationally. But if you don't have the clinicians to follow up, to know what to do properly, then that's a problem, that's a huge problem. I think that really needs to be addressed right now too and I don't know what the answer is with that because I know [a clinical geneticist] tells me, and I hear it everywhere, that there's a real shortage in physicians who really want to specialize in the field and I don't know if it's because of the challenges we do have trying to do our jobs [or] because of the limitations we have because we're not a big money-making clinic. A lot of nutritionists wouldn't really go into this field at all, given that there's a lot more. It's just a different focus from say a diabetes counseling session. I think you just really have to like this field otherwise you can get overwhelmed by it. (B0111)*

### *Education Concerns*

Education was another concern when considering NBS expansion. Similar to concerns about education about current conditions, there was concern that education both within and outside of the field needed to be improved. As one interviewee described, conditions with low severity also cause problems for families:

*The only other...barrier is when we get these screen positive children but what they're positive for is more of a benign condition in the end and helping the family really wrap their heads around that because they see their child as having a disease. So I think there's a challenge in... you ramp these parents up that they might have something. That seems to be an increasing challenge because we're picking up more and more children with these benign variants, so that's definitely a new challenge and I'm not sure what the best resource or solution is for those groups yet. (C0317)*

Providers also described the need to educate workers within the field about the conditions (B0313, A043), including themselves (B0313) and general geneticists (A043). One nutritionist discussed the need for, and difficulty of, treatment protocol verification (B0111):

*I think there needs to be a lot more treatment protocol clarification, like for a particular rare disorder, you know, what is the most appropriate, what is the most effective, evidence-based effective treatment for them, medical or nutritional. What is the benefit to them for some of the really, really rare disorders, of finding them and then starting their treatment early. Is that helpful or not, that kind of thing, getting them to the referrals they need, whether it's neurology as well as metabolism? These are so rare that we don't have a good base of studies of good RCT, randomized controlled trials that can tell us...what we should be doing with a lot of these really rare disorders. (B0111)*

### *Ethical Concerns*

Interviewees also expressed ethical concerns about NBS expansion. One nutritionist expressed encouragement at being able to find children who otherwise would not have received a diagnosis while also expressing concern about the existing level of knowledge surrounding treatments for these conditions

(B0111). Clinical geneticists were also concerned about treatment (A027, A0510) and why the new conditions were being added (A027).

*Well, certainly, I think in a good way that we'll hopefully find all those children out there who might have been missed otherwise, who would be clinically hurt because of it, because of a late diagnosis...I think it's an exciting thing but I think there's a lot of work that still needs to be done to clarify... what the point of it is, what the goal [is], what the treatment [is]. (B0111)*

*I think however, though, it has to be really weighed, because...I don't think that just because we can, we should. And that goes to the fact that if it can't be treated, if it's going to give more false positives, if the negatives and the true positives are not truly going to be symptomatic, you know all those kinds of things, I think it has to be strongly considered because it will weigh on because you'll get much more referrals, you'll be getting much more work, and you'll be stressing out much more families and possibly for nothing. And it's one of the things that I do hate about some conditions like VLCAD, where...we've kind of gotten into this conundrum now where we stuck [the patient] out onto this 6-8 week process no matter what almost. And I hate that, because I know the majority are not going to be effected and for 6-8 weeks I've disrupted this family's life. (A027)*

*These diseases are going to raise questions about follow-up, treatment, and confirmation that frankly we haven't dealt with up until now. They're quite difficult questions for which there are no clear answers at the current time. (A0510)*

**Next Steps:**

This study was conducted in one region of the United States, the northeast. And while there is a high prevalence comparatively of metabolic cases here, it is obviously different than other areas of the country in terms of both resources and patient mix. What was evidenced here relative to the variation in care and coordination of care should be evidenced in other areas, if such the drivers of such variation are hypothesized to be different. If not, the question arises as to whether resources devoted to caring for metabolic patients should be measured more statically and statistically to provide a benchmark of service capacity versus demand. Doing so would enable better estimations of what provider capacity ought to be relative to population and condition prevalence, as well as to estimate changes in provider supply needs given changes and advances in the fields' services and scope.

Understanding resource use will further help inform how best to realign resources for the benefit of providers, patients and families. It is also imperative to understand what resources are currently being used for any cost-effectiveness analysis to have meaning. This could be accomplished through broad based surveying if in fact the drivers of resource use and their disparities are believed to be similar through the regions of the country, or continued interviewing if they are thought to differ.

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## Appendix A. Clinician Interview Guide

Thank you for agreeing to participate in this interview. My name is \_\_\_\_ and I am an interviewer for the New England Genetics Workforce Projection Project.

Before we begin, I would like to review some information with you about the interview process. During the interview, I will be asking you a number of questions about newborn screening genetic services. Our study uses a broad definition of genetic services that includes any activity to evaluate, counsel, manage, treat, or coordinate care for an individual or family with a condition that may be inherited or have a genetic basis. In general, this interview takes \_\_\_\_ to \_\_\_\_ minutes.

I would also like to review that you were sent an informed consent form to complete acknowledging your agreement to participate in the phone interview. Do you consent to participate? YES/NO. Do you consent to my making an audiotape of our interview, which will only be used for research purposes to allow us to review and clarify any responses? YES/NO. If any of the questions make you uncomfortable please let me know; you will not be obligated to answer them. Do you have any questions before we begin? YES/NO.

### **Interview Set-Up**

Individual Interviewed (name):

Date of Interview:

Organizational Affiliation of Interviewee if available:

Interviewed by:

Time of Interview: Begin:                      End:

Duration of Interview:

Transcription Time:

### **CORE QUESTION:**

I am going to be asking you a set of questions about patients with metabolic conditions detected by newborn screening that are commonly seen in your practice. I'd like to begin by discussing patients with "XYZ disease."

*Note to interviewer: This disease should be taken from the ranked list question of the clinic survey for the associated clinic for this provider*

**Q. A.1: Please follow a typical patient for your practice with "XYZ" through the continuum of care you provide from the time of positive newborn screen until his/her first birthday.**

*Note to Interviewer:* Please probe in particular about "work" related to the urgency of first contact on a positive screen, the extent of the initial diagnostic work-up, and the complexity of disease management

*Note to Interviewer: Because we expect the interviewee to be giving you a narrative, you may or may not need to ask these questions. However, by the end of the interview we will want to have obtained this information:*

- What is your role in the care of this patient?
- How much of your time does each of these activities take?
  - How much time do you spend on the phone with the family, with the pediatrician (*if not PCP*), with other health care professionals in between clinic visits for this patient?
  - How much time do you spend doing case management for this patient?
  - How much time do you spend talking to insurance companies or doing other administrative tasks for this patient?

*\*\*These responses should include face-to-face time as well as time in between visits.*

- Who else is involved in the patient's care?
- Which providers most often refer metabolic patients to you? (*probe for dietitians, genetic counselors, PCPs, any others if not provided*)
- To which providers do you refer most metabolic patients? (*probe for dietitians, genetic counselors, PCPs, and any others if not provided*)

**Q. A. 2: Administrative Time:**

- Please estimate the amount of administrative time (scheduling, paperwork, referrals) these patients require on average, from time of initial positive screen to age 1 year.

**Q. A. 3: Visit Frequency and Length:**

- Please estimate the number of visits, on average, that patients with XYZ condition require from time of initial positive screen through age one year.
- How frequently do these patients need to be seen?
- Typically, how long is an initial visit?
- Typically, how long is a follow up visit?

**Q.A. 4: Laboratory Testing**

- Do patients with XYZ condition receive any additional testing to confirm the diagnosis? Please specify.

- Do patient with XYZ condition require any other testing during the first year of life? Please specify.

Use these as probes, if not already addressed:

- o IF YES,, How many additional tests do they need?
- o Can this testing all be done in the same visit?
- o How long are visits for these tests?

**Q. B. Are there patients in your practice with other conditions detected on newborn screening that are more or less labor-intensive? Please highlight differences.**

*Notes to Interviewer: 1) Make sure the interviewee gives you the name of the condition(s) being described as more or less labor-intensive. 2) Probe in particular about “work” related to the urgency of first contact on a positive screen, the extent of the initial diagnostic work-up, and the complexity of disease management. Use the previous section as a guide, revisiting the core components*

**Q. C. 1 Barriers or Challenges to Providing Care**

- Please describe any barriers or challenges that you face in providing care to metabolic patients.

*Note to the interviewer: Use the below probes if needed*

- Are there any resources that would help you do your job better?
- Do you have any suggestions for improving quality of care?

**Q. D. 1 Education and Care Coordination**

Next, I am going to ask questions concerning education and care coordination about the representative patient with XYZ condition you’ve mentioned.

- Are there points in the care process that you feel that education about metabolic conditions and their care could be enhanced? (*probe for where / who / how*) To whom? By whom?

**Q. E. 1: Other**

- Is there anything else involved in the treatment of patients with metabolic disorders that you think we should know about?

*If Yes: Probe*

*If No: Conclude call*

**Interview Conclusion**

THANK YOU FOR YOUR PARTICIPATION.

**Health Professional Specific Probes Reference Sheet:**

*Note to the interviewer: Use the below probes if needed when talking with these specific individuals relative to core questions A and B if not touched on in their general responses.*

## **Appendix B: NBS Interview Guide**

### **Newborn Screening Coordinator Interviews**

Thank you for agreeing to participate in this interview. My name is \_\_\_\_ and I am an interviewer for the New England Genetics Workforce Projection Project.

Before we begin, I would like to review some information with you about the interview process. During the interview, I will be asking you a number of questions about newborn screening genetic services. Our study uses a broad definition of genetic services that includes any activity, including genetic testing, to evaluate, counsel, manage, or plan for an individual or family with a condition that may be inherited or have a genetic basis. In general, this interview takes \_\_\_\_ to \_\_\_\_ minutes.

I would also like to review that you were sent an informed consent form to complete acknowledging your agreement to participate in the phone interview. You have indicated that you consent to participate. Is this still accurate? YES/NO You also consented to this interview being recorded for research purposes? YES/NO. As stated in the email you received, you will receive a transcript of this interview in order to clarify or correct any of the information that we discuss. If any of the questions make you uncomfortable please let me know; you will not be obligated to answer them. Do you have any questions before we begin? YES/NO.

### **Interview Set-Up**

Individual Interviewed (name):

Date of Interview:

Interviewed by:

Time of Interview: Begin:                      End:

Duration of Interview:

Transcription Time:

### **Core Questions**

I AM GOING TO ASK YOU QUESTIONS CONCERNING WHAT HAPPENS IN YOUR STATE ONCE A CHILD HAS SCREENED POSITIVE FOR A METABOLIC CONDITION DURING A NEWBORN SCREENING. PLEASE FOLLOW AN AVERAGE PATIENT FROM NEWBORN SCREENING TO ONE YEAR OF TREATMENT, IF TREATMENT IS NECESSARY.

PLEASE EXPLAIN TO ME WHAT HAPPENS ONCE A CHILD HAS SCREENED POSITIVE FOR A METABOLIC CONDITION DURING A NEWBORN SCREEN.

### **INTERVIEW CONCLUSION**

THANK YOU FOR YOUR PARTICIPATION.

BEFORE WE END THE INTERVIEW, IS THERE ANYTHING ELSE ABOUT TREATING PATIENTS WITH METABOLIC CONDITIONS THAT WE SHOULD KNOW ABOUT?

*If Yes: Probe*

*If No: End Call*

## Appendix C: Consent Forms

### THE NEW ENGLAND GENETICS WORKFORCE PROJECT

Dear **(enter name here)**:

The New England Genetics Collaborative at the University of New Hampshire in conjunction with the American College of Medical Genetics is conducting a research project to estimate the projected demand for genetic care services in New England over the next ten years. I am writing to invite you to participate in this project.

If you agree to participate in this study, we will be contacting you by phone and asking you to describe the process of and resources needed to treat two to three example children who screen positive for metabolic conditions at birth. Interviews are anticipated to take approximately 45 to 60 minutes and will be recorded. After the interview you will receive notes taken from your interview, at which time you can make any changes or clarifications you deem necessary.

The attached document is an informed consent form required for participation. It contains information on the study and contact information if you have question regarding the study.

If you would like to participate, **please respond to this email with the most convenient date and time to contact you.**

Sincerely,

Robert J. McGrath PhD  
Assistant Professor  
Department of Health Management and Policy  
University of New Hampshire

**INSTITUTIONAL REVIEW BOARD FOR THE PROTECTION OF HUMAN SUBJECTS IN RESEARCH**

Informed Consent:

Thank you for considering participation in this study. As stated in the introductory email you received, you agree to participate in this study, we will be contacting you by phone and asking you to describe the process of and resources needed to treat two to three example children who screen positive for metabolic conditions at birth. Interviews are anticipated to take approximately 45 to 60 minutes and will be recorded. After the interview you will receive notes taken from your interview, at which time you can make any changes or clarifications you deem necessary.

The potential risks of participating in this study are minimal. The information you provide will be reported anonymously. However, although we intend to maintain your anonymity, we cannot ensure confidentiality because of the small size of the newborn screening and genetics workforce in New England. Although you are not anticipated to receive any direct benefits from participating in this study, the benefits of the knowledge gained will inform our research in estimating resource allocation and infrastructure demand that result from positive newborn metabolic screens.

Participation is strictly voluntary; refusal to participate will involve no prejudice, penalty, or loss of benefits to which you would otherwise be entitled. If you agree to participate and then change your mind, you may withdraw at any time during the study without penalty. Audio data recordings will be kept in a secure data file on my desktop. Interview notes will be kept on a secure computer drive accessible only to members of the research team.

You will not receive any compensation to participate in this project.

If you have any questions about this research project or would like more information before, during, or after the study, you may contact us at 603-862-2962 or at [Robert.McGrath@unh.edu](mailto:Robert.McGrath@unh.edu). If you have questions about your rights as a research subject, you may contact Julie Simpson in the UNH Office of Sponsored Research at 603-862-2003 or [Julie.simpson@unh.edu](mailto:Julie.simpson@unh.edu) to discuss them.

Please fill out the information below and either return it in the envelope provided, or scan and email it to Monica McClain at [monica.mcclain@unh.edu](mailto:monica.mcclain@unh.edu). Please keep a copy for your records.

Yes, I, \_\_\_\_\_ consent/agree to participate in this research project.

No, I, \_\_\_\_\_ do not consent/agree to participate in this research project.

\_\_\_\_\_  
Signature

\_\_\_\_\_  
Date