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A National Assessment of the Newborn Screening Workforce for Metabolic Conditions, Phase Two Report

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Phase Two 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)

negc THE NEW ENGLAND GENETICS COLLABORATIVE

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A National Assessment of the Newborn Screening Workforce

Executive Summary

The findings from this survey provide broad reinforcement to the themes expressed in the first phase interview project conducted in the Northeast region. Overall, providers report that non-care related time spent with patients and on education and administrative tasks is substantial and challenging. Mentioned most frequently (64% of providers) was time spent with insurers for approvals and lacking reimbursements for medical foods and formulas.

Patient care management and education was expressed as time consuming and often as a role taken on by providers other than medical geneticists. Non-medical geneticists were more likely to report spending up to four hours monthly on the phone with families, and up to four hours on non-care related time monthly not insurance related. This becomes important in that when asked what their primary responsibility is, most providers cited patient care, however they spent a large portion of time on non-care activities, mostly non-reimbursed.

Care coordination also varied. Most respondents cited the use of care teams, but composition of those teams varied and roles were not understood. Less than 5% of providers cited communication with primary care providers (PCPs) or other medical staff as primary to their responsibilities. This is despite a majority of providers reporting that PCPs should participate in care teams but often lack knowledge of the conditions. Others reported the need to expand care teams to ease Geneticist workload and compensate for provider shortages. Remote patient monitoring of some kind was also mentioned by 84% of respondents, specifically to deal with geographic remoteness of patients.

Also consistently mentioned was the lack of best practices and care guidelines, especially relative to expanding the NBS panel. While providers remain sheepishly optimistic about expansions, they also expressed concern that the current challenges to providing care will be exacerbated.

Overall a majority of respondents report having substantial work commitments to treating patients with metabolic disorders. Taken together, their responses reflect an uncoordinated system of care, primarily related to extra-care activities. Care process for medically treating patients was quite consistent overall. They are also reporting being undervalued but committed to the work of treating these patients. Many reflected on the need to better integrate care, but lacked incentives or time to do so.

Expansions to the NBS panel will likely not be well met if many of the administrative and educational concerns are not addressed. Further, the field of metabolic genetic medicine is not well understood by the U.S. health care system more broadly. With reimbursement lacking and care coordination a large part of national efforts to curb health spending, addressing the cost effectiveness of providing care to these patients will be paramount. Developing more consistent and effective care teams and care process guidelines should be investigated. This will include consistent measuring of care coordination time and effort to maximize reimbursement from insurers.

A National Assessment of the Newborn Screening Workforce

Overview

This study proposed to assess the scope and intensity of services needed to provide quality genetic health care to newborns diagnosed with metabolic conditions following a positive newborn screen (NBS) from birth to age one across the United States. The resources examined included time spent on face-to-face visits as well as non-visit time for health care providers treating these patients. The term 'health care providers' was broadly defined to include any person who works with the patient in an official health care capacity including clinical geneticists, dietitians/nutritionists, genetic counselors, patient advocates, and office/administrative staff. Previous work on this topic in the New England region has shown that providing care to patients with metabolic conditions requires frequent visits and many non-visit hours of work. [19] Additionally, providers often take on responsibilities that are tangential to their official duties, for example, dietitians often act as care coordinators for their patients and families. This project was designed based on the work in the New England region to determine if these patterns were evident nationally.

Background and Rationale

Newborn screening (NBS) is a state-based universally mandated core public health function that identifies the presence of specific disorders at birth so that treatment can begin before clinical symptoms present, often mitigating adverse health outcomes or death. Most of these conditions are inborn errors of metabolism, and therefore genetic. With improved technologies the number of metabolic conditions tested on the NBS panel continues to expand. However, these expansions highlight three pervasive issues within the current medical genetics workforce: (1) the workforce is inadequate to meet current demand for genetics services; (2) the metabolic conditions screened for on the NBS panel often require high intensity management; and (3) most are rare diagnoses requiring coordinated specialty care.

Researchers studying the medical genetics workforce have concluded that the size of the current clinical genetics workforce[1,2] and the number of students entering the field[2,3] are inadequate to meet the growing demand for genetic services. Clinical geneticists are poorly dispersed across states[1] and a high proportion of geneticists practice in urban areas and academic medical centers,[1,4, 5, 6] that are not accessible to large portions of the population who need services but live a distance away. Clinical geneticists often work with a team of health care providers, including genetic counselors, whose workforce issues must also be addressed and understood.[1,4,5,7,8]

One commonly cited reason for problems within the medical genetics workforce relates to income and reimbursement. Reimbursement rates for genetic services are poor[1,7] and clinical geneticists report low satisfaction with income and earning potential despite the fact that many clinical geneticists are reporting increases in patient volume[1,5] and complexity.[1] Combined with high levels of labor intensity and job-related stress, beginning a career in medical genetics is reported to be less attractive to future practitioners at a time when the demand for genetic services is growing. [1,2,9]

Workforce capacity is an especially salient issue for NBS because of the intensity of treatment. Visits for new and follow-up patients at genetics clinics for the metabolic conditions on the panel are lengthy, averaging 40 to 65 minutes.[1,5] Genetics providers also spend substantial amounts of time on

patient care activities outside of the clinic visit: approximately three hours on average for new patient visits and two additional hours for follow-up visits.[4,9, 10]

The labor intensity of diagnosing and treating conditions detected on NBS is counterbalanced by two issues. First, according to published research, approximately 2.4 of every 1,000 live births resulted in the detection of a condition identified on NBS in the U.S. in 2010.[11] Very few children actually need treatment; thus, it is difficult to estimate the size of the workforce needed to treat these conditions.[12] Second, while more conditions that have severe health and developmental consequences can be detected by tandem mass spectrometry, [13, 14] there is little to no information about the treatment or natural course of many of these conditions.[9,15, 16, 17] In particular, Steiner reports that there is little evidence-base on the treatment of inborn errors of metabolism because there are few research subjects, most of these diagnoses are made at birth, and treatments may need to be individualized to patient-specific characteristics.[15] Additionally, a national infrastructure for collecting this information was only recently developed (the NBS Translational Research Network (NBSTRN)).[18]

Finally, the growth of genetic medicine is changing the relationships between primary care providers, geneticists, and patients. Relationships between genetic specialists and primary care providers would seem especially important for patients who screen positive for a metabolic condition on NBS.[8] Yet studies have found that most primary care providers' knowledge about genetics remains inadequate.[6-8, 17]

NBS brings to the forefront the tenuous balance between the adequacy of the medical genetics workforce and the rare yet labor-intensive conditions screened for on the NBS panel.

Because research to date has not directly studied the workforce needed to care for children diagnosed with a metabolic condition during NBS, a first phase of empirical study into this issue was conducted by the New England Genetics Collaborative at the University of New Hampshire for the NCC to examine issues of care resources being utilized for metabolic patients, as well as extra-care activities such as education with families, administrative tasks, and care coordination between providers.[19] That study examined the process of care for children with a positive metabolic newborn screen through expert interviews with providers across the New England Region. Findings suggested that the NBS process worked well in the states, but that resource intensity varied markedly depending on the condition with which the child was diagnosed. Also found was that care coordination was lacking both between treating providers and primary care providers and the metabolic team, and that the roles of care teams were often diverse and not well understood or communicated. Educational and administrative burden was also found to be substantial. Many providers believed that while there is potentially great promise from enhanced metabolic screening, the workload and process issues that currently exist would be exacerbated.[19]

These findings provide an important empirical window into the care process for metabolic patients broadly defined. It was, however, conducted on a limited geographic sample of providers in the Northeast. Further data collection was warranted to understand if similar issues were being experienced in other parts of the country. For this reason, a larger national survey was developed as a second phase of this work, informed from the findings of the first.

Methodology

Similar to phase one, the phase two survey sought to define the process of care for newborns diagnosed with metabolic conditions after positive newborn screens (NBS) from birth to age one based on reports from a national sample of health care providers who treat such patients. Metabolic conditions were selected because they provide a more definable treatment protocol than some other disorders on the NBS panel and thus allow for more accurate empirical measurement of the resources needed to treat those patients. Additionally, it examined the amount of time that providers spent working on these patients' cases outside of visit time, potential challenges to providing care, and attitudes about an expanded NBS panel. These topics were defined based on the phase one interview project conducted during 2010.

In order to assess these factors at the national level, an online survey was created. In addition to the phase one findings, input into the survey was sought from the regional collaboratives nationwide. Those in the West and Midwest cited the use of telemedicine and access as additional areas of interest for inclusion in the survey. The final tool, found in Appendix A, contained seventy-four questions and was deployed by the University of New Hampshire Survey Center. The survey opened on September 28, 2011 and closed on May 1, 2012. Participants were recruited in two stages. First, potential participants were contacted through the Regional Genetic and Newborn Screening Service Collaboratives. Second, participants were recruited via professional organizations, conferences, and networks. Potential participants were contacted electronically by the National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives (NCC), the Society for Inherited Metabolic Disorders (SIMD), the NBS mailing list (Newborn Screening Inquiry/Discussion List) by Dr. Brad Therrell (NBS@lists.UTSCSA.edu), and the Pediatric and Clinical Genetics special interest group listserv of the National Society of Genetic Counselors (NSGC). Finally, the survey was made available electronically and in paper format at the American College of Medical Genetics (ACMG) Annual Clinical Genetics Meetings (March 27-31, 2012) and the Genetic Metabolic Dietitians International (GMDI) Educational Conference (April 19-21, 2012). The sampling methodology is detailed in Appendix B.

Analyses were conducted using Stata 11. The analyses created for this report are based on the 114 usable surveys that were submitted. Sample sizes vary for each section of the survey because of skip patterns and missing data; respondents who responded to at least one question in a given section were included in that section's analysis. The final sample contained 67 clinical geneticists and 47 other (non-clinical geneticist) providers. Process of care and non-visit administrative time analyses were calculated separately for clinical geneticists and all other providers. Significance tests were not calculated because of thin cells. Providers were invited to write-in comments about 'best practices' and other concerns not covered by the survey at the end of the survey. These comments have been coded thematically and incorporated with statistical findings.

Summary of Findings

The sections below represent a summary of the key findings from each area of the survey. Detailed analyses are presented in the following section.

Care Process

The intensity of services provided to newborns with metabolic conditions detected on NBS can be described by the frequency, length, and duration of contact between patients/families and providers.

- Providers are in contact – via telephone and face-to-face visits – with patients and their families very soon after a metabolic condition is detected on NBS.
- Providers are in contact with patients frequently during the first year of life. Although metabolic disorders vary, providers seem to have frequent contact with patients who have any type of metabolic disorder.
- Providers see patients less frequently as patients get older during the first year of life.

Clinical geneticists and other providers have similar rates of contact with providers for most disorder categories. However, for some metabolic conditions, for example, amino acid disorders, other providers reported more frequent visits with patients/families than did clinical geneticists.

Non-Visit Administrative Time

Administrative tasks, such as talking on the phone with families, doing case management, interacting with insurance companies, and preparing for face-to-face visits, were reported as time-consuming tasks.

Differences were evident by provider type: other providers more frequently reported spending more time on the phone with families, doing case management, preparing for face-to-face visits, working with insurance companies, and on other tasks than did clinical geneticists.

The most time-intensive reasons that providers reported were related to insurance; specifically approvals for medical foods and formulas, approvals for medications, and general reimbursement for services.

Care Coordination and Models of Care Provision

Providers reported many responsibilities for patients. Providers most frequently reported being responsible for managing medical needs and educating patients and families. Other providers frequently reported coordinating care for patients and families.

Different models of care provision were used by providers' clinics to address patient needs; most important was using a team approach to care. Providers also frequently reported using satellite clinics and using telemedicine technologies.

Challenges to Providing Care

Practitioners reported that their ability to provide care during the first year of life is primarily made challenging by the lack of reimbursement for patient care. Other factors, such as parental/familial knowledge and awareness of the disorder, compliance with treatment and management protocols were considered somewhat challenging.

However, some factors were considered less challenging. Specifically, providers' contact with state NBS or follow-up programs, the ability to transition patients to hospital providers, and the practitioners' own educational needs concerning metabolic disorders were not considered challenging.

Education

Providers reported spending substantial amounts of time educating families about metabolic conditions. Families needed to be educated about the treatment and management of the disorders in addition to other basic information.

Providers mainly used pamphlets, resource binders, and websites to educate their patients and families. Families, providers reported, only sometimes had accurate information about metabolic disorders when they came into the provider's practice.

Relationships with Primary Care Providers

Metabolic providers reported that relationships with primary care providers were important and that primary care providers should participate in the care of patients with metabolic conditions, despite what was perceived as a lack of knowledge about these conditions.

Attitudes about Expanding the Newborn Screening Panel

Providers seemed cautiously optimistic about expanding the NBS panel. While they were excited to treat children whose conditions would otherwise go undetected, they remain concerned about the number of trained providers, the ability to spend enough time with new patients, and about being reimbursed for care provided.

Detailed Analysis and Findings

Detailed analytic findings are presented below. They have been divided topically for clarity. Sample sizes for analyses are indicated.

1. Sample

The final sample contained a total of 114 cases with usable data, as shown in Table 1. The majority of respondents were clinical geneticists (59%). Because one of the goals of this project was to assess processes of care for all providers, the sample was divided based on provider type: clinical geneticists versus all other providers. The majority of other providers were dietitians/nutritionists (38%), genetic counselors (21%), and 'other' providers (21%). These 'other' providers include nurse practitioners, neurologists, and lab directors, among others.

The majority of providers (60%) stated that less than 40% of their practice was dedicated to treating children diagnosed with metabolic conditions following a positive NBS from birth to age one. Eleven percent reported that at least 90% of their practice was composed of these children. Nearly two-thirds of clinical geneticists (63%) reported that less than 30% of their practice was composed of these children, compared to 28% of other providers. Other providers were most likely (26%) to report that at

least 90% of their practice was dedicated to treating children diagnosed with metabolic conditions following a positive NBS.

At least 90% of all providers reported treating children with each of the three types of metabolic disorders on the NBS panel – organic acid disorders (90%), fatty acid oxidation disorders (94%), amino acid disorders (93%). Eighty-nine percent of all providers reported treating children with all of these types of disorders. Clinical geneticists were most likely to report that they treated children with fatty acid oxidation disorders (97%), while other providers were most likely to report that they treated children with amino acid disorders (92%).

Respondents were asked to provide their zip code so that processes of care could be assessed regionally. Most providers (33%) did not report their zip codes and could thus not be assigned to a region. However, providers who were able to be assigned to a region were spread rather evenly across the United States. This remained consistent for clinical geneticists and other providers.

Table 1. Respondent Characteristics[1]

		Provider Type	
	Total Sample (n=114)	Clinical Geneticists (n=67)	Other Providers (n=47)
Provider Type #1			
Clinical Geneticist	58.77% (67)	100% (67)	-
Dietitian/Nutritionist	15.79 (18)	-	38.30 (18)
Social Worker	0.88 (1)	-	2.13 (1)
Nurse	2.65 (3)	-	6.38 (3)
Patient Advocate	0.88 (1)	-	2.13 (1)
Office/Administrative Manager	2.63 (3)	-	6.38 (3)
Genetic Counselor	8.77 (10)	-	21.28 (10)
Primary Care Provider	0.88 (1)	-	2.13 (1)
Other	8.77 (10)	-	21.28 (10)
Percentage of Practice Dedicated to These Children			
0-10%	12.28 (14)	17.91 (12)	4.26 (2)
11-20%	21.05 (24)	23.88 (16)	17.02 (8)
21-30%	14.91 (17)	20.90 (14)	6.38 (3)
31-40%	11.40 (13)	11.94 (8)	10.64 (5)
41-50%	6.14 (7)	5.97 (4)	6.38 (3)
51-60%	7.02 (8)	7.46 (5)	6.38 (3)
61-70%	4.39 (5)	5.97 (4)	2.13 (1)
71-80%	1.75 (2)	1.49 (1)	2.13 (1)
81-90%	4.39 (5)	1.49 (1)	8.51 (4)
91-100%	11.40 (13)	1.49 (1)	25.53 (12)
Not Reported	5.26 (6)	1.49 (1)	10.64 (5)
Providers Treat Children Diagnosed with…			
Organic Acid Disorders	90.35 (103)	94.03 (63)	85.11 (40)
Fatty Acid Oxidation Disorders	93.86 (107)	97.01 (65)	89.36 (42)
Amino Acid Disorders	92.98 (106)	94.03 (63)	91.49 (43)
U.S. Census Region			

Northeast	17.54 (20)	17.91 (12)	17.02 (8)
South	17.54 (20)	17.91 (12)	17.02 (8)
Midwest	14.04 (16)	10.45 (7)	19.15 (9)
West	18.42 (21)	19.40 (13)	17.02 (8)
Not Reported	32.46 (37)	34.33 (23)	29.79 (14)

[1]Column percentages are shown.

2. Processes of Care

In order to assess the scope and intensity of services provided to patients, this project distinguished between three categories of metabolic disorders: organic acid disorders, fatty acid oxidation disorders, and amino acid disorders. While there is substantial variation within each category of disorder, these categories serve to identify the different disorders and treatment protocols appropriate to each type of disorder. Additionally, as one provider stated in the Best Practices section of the survey, processes of care vary by "unique factors (insurance, level of education/understanding, geographic location, etc.)" it can be difficult to "generalize a specific plan to all patients." The following analyses identify the disorder category and associated process of care. For each category of disorder, providers were asked to indicate the individual disorders they saw most commonly in their practice and the length and frequency of initial and follow-up visits and phone calls.

Organic Acid Disorders

Among the 103 respondents who reported that they treated children diagnosed with organic acid disorders following positive NBS, only 94 answered any questions regarding the specific conditions they saw or services they provided. The results presented below reflect the reports of those 94 respondents.

As shown in Table 2a, the most frequently reported organic acid disorders seen in practice were Priopionic Academia (PROP; 66%) and Methylmalonic Acidemia (Methylmalonyl-CoA Mutase) (MUT; 62%). Providers also frequently reported 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC; 42%), Glutaric Acidemia Type I (GA1; 40%), and Methylmalonic Acidemia (Cobalamin Disorders) (Cbl, A, B; 37%). This pattern remained the same for clinical geneticists. Among other providers, Propionic Acidemia (PROP; 68%) was the most frequently reported disorder, followed by Methylmalonic Acidemia (Methylmalonyl-CoA Mutase) (MUT; 50%) and Isovaleric Acidemia (IVA; 42%).

Table 2a. Organic Acid Disorders Most Often Seen in Practice

		Provider Type	
	Total (n=94)	Clinical Geneticists (n=56)	Other Providers (n=38)
Propionic Academia (PROP)	65.96 (62)	64.29 (36)	68.42 (26)
Methylmalonic Acidemia (Methylmalonyl-CoA Mutase) (MUT)	61.70 (58)	69.64 (39)	50.00 (19)
Methylmalonic Acidemia (Cobalamin Disorders) (Cbl, A, B)	37.23 (35)	37.50 (21)	36.84 (14)
Isovaleric Acidemia (IVA)	29.79 (28)	21.43 (12)	42.11 (16)
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	41.49 (39)	42.86 (24)	39.47 (15)
3-Hydroxy-3-Methyglutaric Aciduria (HMG)	2.13 (2)	3.57 (2)	0.00 (0)
Holocarboxylase Synthase Deficiency (MCD)	3.19 (3)	3.57 (2)	2.63 (1)

B-Ketothiolase Deficiency (BKT)	3.19 (3)	1.79 (1)	5.26 (2)
Glutaric Acidemia Type I (GA1)	40.43 (38)	44.64 (25)	35.21 (13)

Table 2b contains providers' reports of the frequency, timing, and duration of visits for children diagnosed with organic acid disorders during the first year of life. More than half of providers reported talking to the patient's family on the phone the same day or day after a positive NBS. An additional 30% make this phone call within one week of the detection of an organic acid disorder. While most clinical geneticists and other providers reported talking on the phone with patients' families within one week, clinical geneticists were more likely than other providers to report that this first phone call occurred the day of or day after positive NBS. This first phone call usually lasts less than one hour (88%).

The first patient visit was also reported to occur quickly. Thirty percent of providers reported seeing patients with positive NBS the same day or the day after an abnormal NBS; an additional 45% reported seeing patients within one week of the abnormal blood result. Clinical geneticists more frequently reported seeing patients sooner (within the week) than other providers, who were most likely to report seeing patients within two weeks (68%) of the abnormal blood result. This first visit was lengthy. More than 75% of providers reported that the first visit lasted between one and two hours; an additional 17% reported that it lasted between two and four hours. The length of the first visit was similar among clinical geneticists and other providers.

After these initial contacts, patients were seen less frequently by providers over the course of the first year of life. During the first month, approximately 60% of providers reported seeing patients between weekly and a few times per month. During months two and three, patients were reported to visit providers between a few times a month (27%) and once a month (37%). During months four through six, providers reported that they saw patients between once a month (37%) and once every couple of months (45%). Finally, during months seven through twelve, 75% of providers reported that they saw patients with organic acid disorders every couple of months. These follow-up visits were also lengthy. Approximately 70% of providers reported that these visits lasted between 30 minutes and 1 hour. The reported frequency and duration of follow-up visits was similar for clinical geneticists and other providers.

Table 2b. Process of Care for Children Treated for Organic Acid Disorders[1]

		Provider Type	
	Total (n=94)	Clinical Geneticists (n=56)	Other Providers (n=38)
First Phone Call			
The Same or Next Day	52.13 (49)	66.07 (37)	31.58 (12)
Within the Week	28.72 (27)	19.64 (11)	42.11 (16)
Within Two Weeks	7.45 (7)	3.57 (2)	13.16 (5)
Within the Month	5.32 (5)	5.36 (3)	5.26 (2)
Within Six Months	1.06 (1)	0.00 (0)	2.63 (1)
Not Reported	5.32 (5)	5.36 (3)	5.26 (2)
Length of First Phone Call			
Less than 1 Hour	88.30 (83)	83.93 (47)	94.74 (36)
1-2 Hours	6.38 (6)	10.71 (6)	0.00 (0)
Not Reported	5.32 (5)	5.36 (3)	5.26 (2)
First Patient Visit			

The Same or Next Day	29.79 (28)	37.50 (21)	18.42 (7)
Within the Week	44.68 (42)	47.43 (26)	42.11 (16)
Within Two Weeks	17.02 (16)	10.71 (6)	26.32 (10)
Within the Month	5.32 (5)	3.57 (2)	7.89 (3)
Within Six Months	1.06 (1)	0.00 (0)	2.63 (1)
Not Reported	2.13 (2)	1.79 (1)	2.63 (1)
Length of First Visit			
Less than 1 Hour	4.26 (4)	5.36 (3)	2.63 (1)
1-2 Hours	76.60 (72)	75.00 (42)	78.95 (30)
2-4 Hours	17.02 (16)	17.86 (10)	15.79 (6)
Not Reported	2.13 (2)	1.79 (1)	2.63 (1)
Frequency of Follow-Up Visits			
First Month			
More than Once a Week	3.19 (3)	3.57 (2)	2.63 (1)
Once a Week	30.85 (29)	32.14 (18)	28.95 (11)
2-3 Times per Month	31.91 (30)	30.36 (17)	34.21 (13)
Once a Month	22.34 (21)	26.79 (15)	15.79 (6)
Every Couple of Months	5.32 (5)	5.36 (3)	5.26 (2)
Never	3.19 (3)	0.00 (0)	7.89 (3)
Not Reported	3.19 (3)	1.79 (1)	5.26 (2)
Months 2-3			
Once a Week	5.32 (5)	3.57 (2)	7.89 (3)
2-3 Times per Month	26.60 (25)	23.21 (13)	31.58 (12)
Once a Month	37.23 (35)	42.86 (24)	28.95 (11)
Every Couple of Months	21.28 (20)	21.43 (12)	21.05 (8)
Never	2.13 (2)	0.00 (0)	5.26 (2)
Not Reported	7.45 (7)	8.93 (5)	5.26 (2)
Months 4-6			
2-3 Times per Month	11.70 (11)	8.93 (5)	15.79 (6)
Once a Month	37.23 (35)	39.29 (22)	34.21 (13)
Every Couple of Months	44.68 (42)	46.43 (26)	42.11 (16)
Not Reported	6.38 (6)	5.36 (3)	7.89 (3)
Months 7-12			
2-3 Times per Month	2.13 (2)	1.79 (1)	2.63 (1)
Once a Month	17.02 (16)	12.50 (7)	23.68 (9)
Every Couple of Months	74.47 (70)	80.36 (45)	65.79 (25)
Not Reported	6.38 (6)	5.36 (3)	7.89 (3)
Length of Follow-Up Visits			
15-30 Minutes	12.77 (12)	16.07 (9)	7.89 (3)
30-45 Minutes	40.43 (38)	33.93 (19)	50.00 (19)
45 Minutes-1 Hour	28.72 (27)	28.57 (16)	28.95 (11)
More than 1 Hour	15.96 (15)	19.64 (11)	10.53 (4)
Not Reported	2.13 (2)	1.79 (1)	2.63 (1)

[1]Column percentages are shown.

Fatty Acid Oxidation Disorders

Among the 107 respondents who reported that they treated children diagnosed with fatty acid oxidation disorders following positive NBS, only 92 answered any questions regarding the specific conditions they saw or services they provided. The results presented below reflect the reports of those 92 respondents.

As shown in Table 2c, the most frequently reported fatty acid oxidation disorders were Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD; 94%) and Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD; 85%). These were also the most commonly reported conditions for clinical geneticists and other providers.

Table 2c. Fatty Acid Oxidation Disorders Most Often Seen in Practice

		Provider Type	
	Total (n=92)	Clinical Geneticists (n=54)	Other Providers (n=38)
Carnitine Uptake Defect/Carnitine Transport Defect (CUD)	50.00 (46)	61.11 (33)	34.21 (13)
Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	93.48 (86)	96.30 (52)	89.47 (34)
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	84.78 (78)	83.33 (45)	86.84 (33)
Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)	34.78 (32)	31.48 (17)	39.47 (15)
Trifunctional Protein Deficiency (TFP)	6.52 (6)	3.70 (2)	10.53 (4)

Table 2d contains providers' reports of the frequency, timing, and duration of visits for children diagnosed with fatty acid oxidation disorders during the first year of life. Nearly half of providers reported talking to the patient's family on the phone the same day or day after a positive NBS. An additional 33% make this phone call within one week of the detection of a fatty acid oxidation disorder. While most clinical geneticists and other providers reported talking on the phone with patients' families within one week, clinical geneticists were more likely than other providers to report that this first phone call occurred the day of or day after positive NBS. This first phone call usually lasts less than one hour (84%); this was similar for clinical geneticists and other providers.

The first patient visit was also reported to occur quickly. Twenty percent of providers reported seeing patients with positive NBS the same day or the day after an abnormal NBS; 46% reported seeing patients within one week of the abnormal blood result; and 26% reported seeing patients within two weeks. The timing of this first visit was similar for clinical geneticists and other providers. This first visit was lengthy. Eighty percent of providers reported that the first visit lasted between one and two hours; an additional 11% reported that it lasted between two and four hours. The length of the first visit was similar among clinical geneticists and other providers.

After these initial contacts, patients were seen less frequently by providers over the course of the first year of life. During the first month, approximately 40% of providers reported seeing patients once a month. During months two and three, patients were reported to visit providers between once a month (32%) and every couple of months (37%). Between months 4 and 12, most providers reported that they saw patients once every couple of months. Most providers (41%) reported that follow-up visits lasted between 30 and 45 minutes, and an additional 30% reported follow-up visits lasting 45 minutes to 1 hour. The reported frequency and duration of follow-up visits was similar for clinical geneticists and other providers.

Table 2d. Process of Care for Children Treated for Fatty Acid Oxidation Disorders[1]			
		Provider Type	
	Total (n=92)	Clinical Geneticists (n=54)	Other Providers (n=38)
First Phone Call			
The Same or Next Day	47.83 (44)	61.11 (33)	28.95 (11)
Within the Week	32.61 (30)	25.93 (14)	42.11 (16)
Within Two Weeks	8.70 (8)	5.56 (3)	13.16 (5)
Within the Month	5.43 (5)	1.85 (1)	10.53 (4)
Not Reported	5.43 (5)	5.56 (3)	5.26 (2)
Length of First Phone Call			
Less than 1 Hour	83.70 (77)	81.48 (44)	89.84 (33)
1-2 Hours	9.78 (9)	12.96 (7)	5.26 (2)
Not Reported	6.52 (6)	5.56 (3)	7.89 (3)
First Patient Visit			
The Same or Next Day	19.57 (18)	22.22 (12)	15.79 (6)
Within the Week	45.65 (42)	48.15 (26)	42.11 (16)
Within Two Weeks	26.09 (24)	24.07 (13)	28.95 (11)
Within the Month	5.43 (5)	3.70 (2)	7.89 (3)
Not Reported	3.26 (3)	1.85 (1)	5.26 (2)
Length of First Visit			
Less than 1 Hour	5.43 (5)	7.41 (4)	2.63 (1)
1-2 Hours	80.43 (74)	77.78 (42)	84.21 (32)
2-4 Hours	10.87 (10)	12.96 (7)	7.89 (3)
Not Reported	3.26 (3)	1.85 (1)	5.26 (2)
Frequency of Follow-Up Visits			
First Month			
More than Once a Week	4.35 (4)	3.70 (2)	5.26 (2)
Once a Week	18.48 (17)	22.22 (12)	13.16 (5)
2-3 Times per Month	21.74 (20)	18.52 (10)	26.32 (10)
Once a Month	40.22 (37)	46.30 (25)	31.58 (12)
Every Couple of Months	9.78 (9)	7.41 (4)	13.16 (5)
Never	1.09 (1)	0.00 (0)	2.63 (1)
Not Reported	4.35 (4)	1.85 (1)	7.89 (3)
Months 2-3			
Once a Week	4.35 (4)	3.70 (2)	5.26 (2)
2-3 Times per Month	15.22 (14)	16.67 (9)	13.16 (5)
Once a Month	31.52 (29)	29.63 (16)	34.21 (13)
Every Couple of Months	36.96 (34)	40.74 (22)	31.58 (12)
Not Reported	11.96 (11)	9.26 (5)	15.79 (6)
Months 4-6			
2-3 Times per Month	5.43 (5)	7.41 (4)	2.63 (1)

Once a Month	29.35 (27)	25.93 (14)	34.21 (13)
Every Couple of Months	58.70 (54)	61.11 (33)	55.26 (21)
Not Reported	6.52 (6)	5.56 (3)	7.89 (3)
Months 7-12			
2-3 Times per Month	1.09 (1)	1.85 (1)	0.00 (0)
Once a Month	7.61 (7)	5.56 (3)	10.53 (4)
Every Couple of Months	84.78 (78)	87.04 (47)	81.58 (31)
Not Reported	6.52 (6)	5.56 (3)	7.89 (3)
Length of Follow-Up Visits			
15-30 Minutes	10.87 (10)	9.26 (5)	13.16 (5)
30-45 Minutes	41.30 (38)	35.19 (19)	50.00 (19)
45 Minutes-1 Hour	30.43 (28)	33.33 (18)	26.32 (10)
More than 1 Hour	13.04 (12)	16.67 (9)	7.89 (3)
Not Reported	4.35 (4)	5.56 (3)	2.63 (1)

[1]Column percentages are shown.

Amino Acid Disorders

Among the 106 respondents who reported that they treated children diagnosed with amino acid disorders following positive NBS, only 89 answered any questions regarding the specific conditions they saw or services they provided. The results presented below reflect the reports of those 89 respondents.

As shown in Table 2e, the most frequently reported amino acid disorder was Classic Phenylketonuria (PKU; 94%). Nearly half of providers also reported frequently treating children with Maple Syrup Urine Disease (MSUD). These findings were similar for clinical geneticists and other providers.

Table 2e. Amino Acid Disorders Most Often Seen in Practice

		Provider Type	
	Total (n=89)	Clinical Geneticists (n=51)	Other Providers (n=38)
Argininosuccinic Aciduria (ASA)	39.33 (35)	41.18 (21)	36.84 (14)
Citrullinemia, Type I (CIT)	38.20 (34)	43.14 (22)	31.58 (12)
Maple Syrup Urine Disease (MSUD)	49.44 (44)	50.98 (26)	47.37 (18)
Homocystinuria (HCY)	34.83 (31)	33.33 (17)	36.84 (14)
Classic Phenylketonuria (PKU)	94.38 (84)	96.08 (49)	92.11 (35)
Tyrosinemia, Type I (TYR I)	14.61 (13)	13.73 (7)	15.79 (6)

Table 2f contains providers' reports of the frequency, timing, and duration of visits for children diagnosed with amino acid disorders during the first year of life. Sixty-three percent of providers reported talking to the patient's family on the phone the same day or day after a positive NBS. An additional 18% make this phone call within one week of the detection of an amino acid disorder. Most clinical geneticists reported talking on the phone with patients' families the day of or after an amino acid disorder was detected; among other providers, 45% reported talking to patients' families the same or next day and 29% reported talking to families within a week of the condition's detection. This first

phone call usually lasts less than one hour (80%); this was similar for clinical geneticists and other providers.

The first patient visit was also reported to occur quickly. Thirty-seven percent of providers reported seeing patients with positive NBS the same day or the day after an abnormal NBS and an additional 44% reported seeing patients within one week of the abnormal blood result. Fifty-one percent of clinical geneticists reported seeing the patient for the first time the same or next day; an additional 39% reported seeing the patient within one week of the abnormal NBS. Other providers saw patients less quickly; 21% of other providers saw patients within two weeks of the detection of the amino acid disorder. This first visit was lengthy. Seventy-three percent of providers reported that the first visit lasted between one and two hours; an additional 21% reported that it lasted between two and four hours. The length of the first visit was similar among clinical geneticists and other providers.

After these initial contacts, patients were seen less frequently by providers over the course of the first year of life. During the first three months, providers reported seeing patients once a month. Between months 4 and 12, most providers reported that they saw patients once every couple of months. The timing of visits differed between clinical geneticists and other providers. While most clinical geneticists reported seeing patients once a week during the first month, (41%), once a month during months two through three (37%), and every couple of months during months four through 12, other providers reported seeing patients more frequently. During the first month, other providers reported seeing patients between once a week (29%) and a few times per month (32%). Other providers also frequently reported seeing patients a few times a month (32%) during months two and three and between once a month (37%) and every couple of months (40%) during months four through six. During months seven through twelve, other providers most frequently reported seeing patients every couple of months (66%). Most providers (46%) reported that follow-up visits lasted between 30 and 45 minutes, and an additional 29% reported follow-up visits lasting 45 minutes to 1 hour. The reported duration of follow-up visits was similar for clinical geneticists and other providers.

Table 2f. Process of Care for Children Treated for Amino Acid Disorders[1]

		Provider Type	
	Total (n=89)	Clinical Geneticists (n=51)	Other Providers (n=38)
First Phone Call			
The Same or Next Day	62.92 (56)	76.47 (39)	44.74 (17)
Within the Week	17.98 (16)	9.80 (5)	28.95 (11)
Within Two Weeks	11.24 (10)	7.84 (4)	15.79 (6)
Within the Month	2.25 (2)	1.96 (1)	2.63 (1)
Not Reported	5.62 (5)	3.92 (2)	7.89 (3)
Length of First Phone Call			
Less than 1 Hour	79.78 (71)	78.43 (40)	81.58 (31)
1-2 Hours	14.61 (13)	17.65 (9)	10.53 (4)
Not Reported	5.62 (5)	3.92 (2)	7.89 (3)
First Patient Visit			
The Same or Next Day	37.08 (33)	50.98 (26)	18.42 (7)
Within the Week	43.82 (39)	39.22 (20)	50.00 (19)
Within Two Weeks	14.61 (13)	9.80 (5)	21.05 (8)
Within the Month	2.25 (2)	0.00 (0)	5.26 (2)

Within Six Months	1.12 (1)	0.00 (0)	2.63 (1)
Not Reported	1.12 (1)	0.00 (0)	2.63 (1)
Length of First Visit			
Less than 1 Hour	4.49 (4)	5.88 (3)	2.63 (1)
1-2 Hours	73.03 (65)	72.55 (37)	73.68 (28)
2-4 Hours	21.35 (19)	21.57 (11)	21.05 (8)
Not Reported	1.12 (1)	0.00 (0)	2.63 (1)
Frequency of Follow-Up Visits			
First Month			
More than Once a Week	5.62 (5)	3.92 (2)	7.89 (3)
Once a Week	35.96 (32)	41.18 (21)	28.95 (11)
2-3 Times per Month	25.84 (23)	21.57 (11)	31.58 (12)
Once a Month	23.60 (21)	29.41 (15)	15.79 (6)
Every Couple of Months	4.49 (4)	3.92 (2)	5.26 (2)
Never	2.25 (2)	0.00 (0)	5.26 (2)
Not Reported	2.25 (2)	0.00 (0)	5.26 (2)
Months 2-3			
Once a Week	11.24 (10)	9.80 (5)	13.16 (5)
2-3 Times per Month	26.97 (24)	23.53 (12)	31.58 (12)
Once a Month	32.58 (29)	37.25 (19)	26.32 (10)
Every Couple of Months	19.10 (17)	19.61 (10)	18.42 (7)
Never	2.25 (2)	0.00 (0)	5.26 (2)
Not Reported	7.87 (7)	9.80 (5)	5.26 (2)
Months 4-6			
Once a Week	3.37 (3)	3.92 (2)	2.63 (1)
2-3 Times per Month	16.85 (15)	19.61 (10)	13.16 (5)
Once a Month	32.58 (29)	29.41 (15)	36.84 (14)
Every Couple of Months	41.57 (37)	43.14 (22)	39.47 (15)
Never	-	-	-
Not Reported	5.62 (5)	3.92 (2)	7.89 (3)
Months 7-12			
2-3 Times per Month	6.74 (6)	9.80 (5)	2.63 (1)
Once a Month	17.98 (16)	13.73 (7)	23.68 (9)
Every Couple of Months	68.54 (61)	70.59 (36)	65.79 (25)
Not Reported	6.74 (6)	5.88 (3)	7.89 (3)
Length of Follow-Up Visits			
15-30 Minutes	4.49 (4)	3.92 (2)	5.26 (2)
30-45 Minutes	46.07 (41)	43.14 (22)	50.00 (19)
45 Minutes-1 Hour	29.21 (26)	27.45 (14)	31.58 (12)
More than 1 Hour	17.98 (16)	23.53 (12)	10.53 (4)
Not Reported	2.25 (2)	1.96 (1)	2.63 (1)

[1]Column percentages are shown.

3. Non-Visit Administrative Time

All providers and office/administrative staff were asked to estimate the amount of time that they personally spent outside of the face-to-face visit doing, for example, case management and preparing for face-to-face visits, per month. These questions were asked once, not separately for the three categories of metabolic disorders identified on NBS described above. The time spent on administrative tasks has been separated based on provider type to reflect the responsibilities of clinical geneticists and other providers. A total of 93 respondents answered at least one question about the amount of time they spent on non-visit administrative tasks.

Table 3a shows the amount of time that providers spent on the administrative tasks. Nearly 30% of providers reported spending less than 30 minutes on the phone with the family of a patient with a metabolic disorder per month. One quarter of providers stated that they spent between thirty minutes and one hour on the phone with families each month, while an additional 22% reported spending between one and two hours on the phone each month. Eight percent of all providers reported that talking to the family on the phone was not one of their administrative responsibilities. Differences in time spent on the telephone were evident by provider type. Clinical geneticists were more likely than other providers to report spending less than 30 minutes on the phone with families while other providers were more likely than clinical geneticists to report spending between two and four hours per month on the phone with families.

Providers reported spending a substantial amount of time on case management each month. Providers most frequently reported spending between thirty minutes and one hour on this task (31%), while only 11% reported spending less than 30 minutes on case management per month. Clinical geneticists most frequently reported spending between thirty minutes and one hour on case management per month, compared to 20% of other providers. Other providers most frequently reported spending more than four hours on this task per month (27%).

Providers most frequently reported spending between thirty minutes and one hour preparing for face-to-face visits each month. Differences in the amount of time spent on this task were evident between clinical geneticists and other providers. Forty-two percent of clinical geneticists reported spending this much time on face-to-face visits. Other providers most frequently reported spending between one and two hours preparing for face-to-face visits (27%).

Providers were also asked how many hours they spent on other, non-specified administrative tasks per month. Providers most frequently reported spending less than thirty minutes or more than four hours on these tasks. Again, differences in the amount of time spent by clinical geneticists and other providers were evident. Clinical geneticists most frequently reported spending less than thirty minutes on other administrative tasks each month (33%). Providers were asked to describe these other administrative tasks. Clinical geneticists descriptions included: documentation and dictation; developing care plans; writing emergency letters; communicating with clinic staff, primary care physicians, other health professionals, and non-parent family members; reviewing lab results; coordinating patient care with insurance companies and regional services; advocating for patients and connecting families with support groups; and working with the NBS program. Comparatively, other providers most frequently reported spending more than four hours on these tasks each month (29%). Tasks reported by other providers included: reviewing lab results and revising diet management plans; documentation; writing emergency letters and letters of medical necessity; communicating with primary care providers, clinical geneticists, and other health care professionals; coordinating patient care; educating families and gathering educational materials; compiling information for newsletters; procuring medical foods and formulas; and connecting families with area resources, including other families whose child(ren) has/have the same disorder.

Table 3a. Non-Visit Administrative Time[1]			
		Provider Type	
	Total (n=93)	Clinical Geneticists (n=52)	Other Providers (n=41)
Time Spent on Phone with Family per month			
Less than 30 minutes	29.03 (27)	34.62 (18)	21.95 (9)
30 minutes-1 hour	24.73 (23)	25.00 (13)	24.39 (10)
1-2 hours	21.51 (20)	23.08 (12)	19.51 (8)
2-4 hours	11.83 (11)	7.69 (4)	17.07 (7)
More than 4 hours	5.38 (5)	3.85 (2)	7.32 (3)
I don't do this	7.53 (7)	5.77 (3)	9.76 (4)
Not Reported	-	-	-
Time Spent Doing Case Management per month			
Less than 30 minutes	10.75 (10)	7.69 (4)	14.63 (6)
30 minutes-1 hour	31.18 (29)	40.38 (21)	19.51 (8)
1-2 hours	19.35 (18)	25.00 (13)	12.20 (5)
2-4 hours	17.20 (16)	13.46 (7)	21.95 (9)
More than 4 hours	18.28 (17)	11.54 (6)	26.83 (11)
I don't do this	3.23 (3)	1.92 (1)	4.88 (2)
Not Reported	-	-	-
Time Spent Preparing for Face-to-Face Visits per month			
Less than 30 minutes	15.05 (14)	17.31 (9)	12.20 (5)
30 minutes-1 hour	31.18 (29)	42.31 (22)	17.07 (7)
1-2 hours	22.58 (21)	19.23 (10)	26.83 (11)
2-4 hours	12.90 (12)	9.62 (5)	17.07 (7)
More than 4 hours	12.90 (12)	7.69 (4)	19.51 (8)
I don't do this	1.08 (1)	0.00 (0)	2.44 (1)
Not Reported	4.30 (4)	3.85 (2)	4.88 (2)
Time Spent on Other Administrative Tasks per month			
Less than 30 minutes	23.66 (22)	32.69 (17)	12.20 (5)
30 minutes-1 hour	17.20 (16)	15.38 (8)	19.51 (8)
1-2 hours	17.20 (16)	15.38 (8)	19.51 (8)
2-4 hours	13.98 (13)	15.38 (8)	12.20 (5)
More than 4 hours	21.51 (20)	15.38 (8)	29.27 (12)
I don't do this	1.08 (1)	1.92 (1)	0.00 (0)
Not Reported	5.38 (5)	3.85 (2)	7.32 (3)
Do You Interact with insurance Companies			
No	27.96 (26)	30.77 (16)	24.39 (10)
Yes	70.97 (66)	67.31 (35)	75.61 (31)
Not Reported	1.08 (1)	1.92 (1)	0.00 (0)

[1]Column percentages are shown.

Providers were also asked whether they interacted with insurance companies. As seen in Table 3a, 71% of providers reported doing so. Other providers more frequently reported interacting with insurance companies than did clinical geneticists. Table 3b contains information on the amount of time that providers worked with insurance companies; this analysis includes only those providers who reported working with insurance companies. Overall, providers most frequently reported spending less than thirty minutes or between one and two hours interacting with insurance companies. Differences were evident by provider type. Clinical geneticists most frequently reported spending less than thirty minutes working with insurance companies (26%), while other providers most frequently reported spending between one and two hours (29%) working with insurance companies.

Table 3b. Amount of Time Spent Interacting with Insurance Companies per month[1]

		Provider Type	
	Total (n=66)	Clinical Geneticists (n=35)	Other Providers (n=31)
Less than 30 minutes	21.21 (14)	25.71 (9)	16.13 (5)
30 minutes -1 hour	18.18 (12)	22.86 (8)	12.90 (4)
1-2 hours	21.21 (14)	14.29 (5)	29.03 (9)
2-4 hours	19.70 (13)	20.00 (7)	19.35 (6)
More than 4 hours	16.67 (11)	14.29 (5)	19.35 (6)
I don't do this	1.52 (1)	0.00 (0)	3.23 (1)
Not Reported	1.52 (1)	2.86 (1)	0.00 (0)

[1]Column percentages are shown.

Providers were also asked to rank the three most time-intensive reasons that they interact with insurance companies. Some providers rated more than one activity in the top three; all of these reports were taken into account in order to describe the variety of reasons why providers interact with insurance companies. These reports are shown in Table 3c. The most time-intensive reason providers reported working with insurance companies was for approvals for medical foods and formulas (64%); this was also the most time-intensive reason reported by both clinical geneticists and other providers. Approvals for medications was reported to be the second most time-intensive reason for working with insurance companies; forty-eight percent of all providers and sixty-two percent of clinical geneticists reported it was second most important. General reimbursement for services was the most frequently reported third most time-intensive reason for interacting with insurance companies. Thirty-six percent of all providers, twenty-seven percent of clinical geneticists, and fifty percent of other providers reported this as the third most time-intensive reasons they worked with insurance companies.

Table 3c. Most Time-Intensive Reasons for Interacting with Insurance Companies among Providers who Interact with Insurance Companies[1]

	First	Second	Third
All Providers (n=66)			
General Reimbursement for Services	9.72 (7)	6.78 (4)	36.07 (22)
Patient Eligibility for Care	8.33 (6)	16.95 (10)	21.31 (13)
Approvals for Medical Foods/Formulas	63.89 (46)	16.95 (10)	4.92 (3)
Approvals for Medications	16.67 (12)	47.46 (28)	6.56 (4)
Approvals for Durable Medical Equipment	0.00 (0)	8.47 (5)	16.39 (10)

Approval for Hospital Services	0.00 (0)	1.69 (1)	8.20 (5)
Other	1.39 (1)	1.69 (1)	6.56 (4)
Clinical Geneticists (n=35)			
General Reimbursement for Services	11.90 (5)	5.88 (2)	27.03 (10)
Patient Eligibility for Care	7.14 (3)	11.76 (4)	21.62 (8)
Approvals for Medical Foods/Formulas	69.05 (29)	8.82 (3)	8.11 (3)
Approvals for Medications	11.90 (5)	61.76 (21)	2.70 (1)
Approvals for Durable Medical Equipment	0.00 (0)	5.88 (2)	21.62 (8)
Approval for Hospital Services	0.00 (0)	2.94 (1)	10.81 (4)
Other	0.00 (0)	2.94 (1)	8.11 (3)
Other Providers (n=31)			
General Reimbursement for Services	6.67 (2)	8.00 (2)	50.00 (12)
Patient Eligibility for Care	10.00 (3)	24.00 (6)	20.83 (5)
Approvals for Medical Foods/Formulas	56.67 (17)	28.00 (7)	0.00 (0)
Approvals for Medications	23.33 (7)	28.00 (7)	12.50 (3)
Approvals for Durable Medical Equipment	0.00 (0)	12.00 (3)	8.33 (2)
Approval for Hospital Services	0.00 (0)	0.00 (0)	4.17 (1)
Other	3.33 (1)	0.00 (0)	4.17 (1)

[1]Column percentages are shown.

Finally, dietitians were asked questions specifically about three-day diet recalls. A total of 17 dietitians completed these questions. As shown in Table 3d, dietitians most frequently reported that 51-60% of their patients and families provide a three-day diet recall (29%). Dietitians most frequently reported spending between fifteen and thirty minutes (53%) preparing a three-day diet recall. Finally, dietitians were asked what their protocol is when patients and families do not provide diet recall records. The majority of dietitians (65%) reported that they complete the diet recall record with the families.

Table 3d. Three Day Diet Recall Protocol among Dietitians

	Total (n=17)
Percent of Patients/Families Providing a 3-day Diet Recall	
0-10%	5.88 (1)
11-20%	-
21-30%	11.76 (2)
31-40%	5.88 (1)
41-50%	11.76 (2)
51-60%	29.41 (5)
61-70%	11.76 (2)
71-80%	17.65 (3)
81-90%	5.88 (1)
91-100%	-
Average Time Spent Preparing a 3-day Diet Recall	
15-30 minutes	11.76 (2)

30-45 minutes	52.94 (9)
45 minutes-1 hour	23.53 (4)
More than 1 hour	5.88 (1)
Not Reported	5.88 (1)
Procedure When Patients/Families Do Not Provide Diet Recall (select all that apply)	
Complete diet recall with them	64.71 (11)
Ask them to complete a diet recall during the visit and review it with them that day	11.76 (2)
Ask them to complete a diet recall at home and send it to the office for review and follow-up	11.76 (2)
Other	11.76 (2)

4. Care Coordination and Models of Care Provision

Providers were asked to answer a number of questions regarding their responsibilities with patients and any models of care provision they used in practice. These questions were asked of all providers; sample sizes vary throughout this section because of skip patterns.

First, providers were asked to rank their top three responsibilities when working with patients and their families. Some providers rated more than one activity in the top three; all of these reports were taken into account in order to describe the variety of responsibilities providers have when working with patients. These are shown in Table 4a. Managing patients' medical needs was the primary responsibility most frequently reported by providers (39%); educating patients and families was the most frequently reported secondary responsibility reported by providers (33%); communicating with primary care providers was the most frequently reported tertiary responsibility reported by providers (23%). Reported responsibilities varied by provider type. Clinical geneticists most frequently reported that they were responsible for managing medical needs (59%), managing dietary needs (37%), and educating patients and families (31%). Other providers most frequently reported managing dietary needs (34%), educating patients and families (42%), and coordinating care for patients and families (26%).

Table 4a. Primary Responsibilities of Providers when Working with Patients and Their Families[1]

	First	Second	Third
All Providers (n=87)			
Coordinate Care for Patient and Family	13.33 (14)	7.37 (7)	17.89 (17)
Interact with Insurance Companies for Patients	0.95 (1)	2.11 (2)	7.37 (7)
Advocate for Patient Services	0.00 (0)	3.16 (3)	6.32 (6)
Manage Dietary Needs	20.00 (21)	24.21 (23)	5.26 (5)
Manage Medical Needs	39.05 (41)	11.58 (11)	3.16 (3)
Educate Patients and Families	16.19 (17)	32.63 (31)	18.95 (18)
Identify/Refer Patients to Community Resources, Support Groups, etc.	0.00 (0)	7.37 (7)	5.26 (5)
Communicate with Primary Care Providers	4.76 (5)	7.37 (7)	23.16 (22)
Communicate with Specialists, Hospital Staff, etc.	2.86 (3)	4.21 (4)	7.37 (7)
Other	2.86 (3)	0.00 (0)	5.26 (5)
Clinical Geneticists (n=49)			
Coordinate Care for Patient and Family	13.11 (8)	5.77 (3)	10.20 (5)

Interact with Insurance Companies for Patients	0.00 (0)	1.92 (1)	2.04 (1)
Advocate for Patient Services	0.00 (0)	3.85 (2)	6.12 (3)
Manage Dietary Needs	9.84 (6)	36.54 (19)	10.20 (5)
Manage Medical Needs	59.02 (36)	13.46 (7)	2.04 (1)
Educate Patients and Families	14.75 (9)	25.00 (13)	30.61 (15)
Identify/Refer Patients to Community Resources, Support Groups, etc.	0.00 (0)	3.85 (2)	2.04 (1)
Communicate with Primary Care Providers	1.64 (1)	7.69 (4)	26.53 (13)
Communicate with Specialists, Hospital Staff, etc.	1.64 (1)	1.92 (1)	6.12 (3)
Other	0.00 (0)	0.00 (0)	4.08 (2)
Other Providers (n=38)			
Coordinate Care for Patient and Family	13.64 (6)	9.30 (4)	26.09 (12)
Interact with Insurance Companies for Patients	2.27 (1)	2.33 (1)	13.04 (6)
Advocate for Patient Services	0.00 (0)	2.33 (1)	6.52 (3)
Manage Dietary Needs	34.09 (15)	9.30 (4)	0.00 (0)
Manage Medical Needs	11.36 (5)	9.30 (4)	4.35 (2)
Educate Patients and Families	18.18 (8)	41.86 (18)	6.52 (3)
Identify/Refer Patients to Community Resources, Support Groups, etc.	0.00 (0)	11.63 (5)	8.70 (4)
Communicate with Primary Care Providers	9.09 (4)	6.98 (3)	19.57 (9)
Communicate with Specialists, Hospital Staff, etc.	4.55 (2)	6.98 (3)	8.70 (4)
Other	6.82 (3)	0.00 (0)	6.52 (3)

[1]Column percentages are shown.

Second, providers were asked about provision of care models used in their practices. Providers were asked to rank from most to least important five models of care provision: using a team approach to care, practicing at satellite clinics, making visits to patient homes, using telemedicine technologies, and serving as a medical home. Providers who reported that their practice used a particular model of care were then asked follow-up questions with regard to that model. A total of eighty-six providers responded to questions regarding models of care.

Overall, ninety-nine percent of providers reported that their clinic used a team approach to caring for patients, forty-five percent reported that they practiced at satellite clinics, nineteen percent reported making home visits, twenty-six percent reported using telemedicine technologies, and sixty-five percent reported serving as a medical home for patients. The comparative importance of models of care is shown in Table 4b. Using a team approach to care was rated as most important by ninety percent of providers. Serving as a medical home and practicing at satellite clinics were most frequently ranked second and third most important models of care provision. Utilizing telemedicine technologies was most frequently ranked fourth most important. Additionally, thirty percent of providers reported that their clinic had a person who worked specifically as a care coordinator (data not shown).

Table 4b. Models of Patient Care Used by All Providers' Practices/Clinics (n=86)[1]

	Most Important	2nd Most Important	3rd Most Important	4th Most Important	Least Important
Using a Team Approach to Care	89.77 (79)	4.76 (3)	2.56 (1)	10.53 (2)	0.00 (0)
Practicing at Satellite Clinics	4.55 (4)	30.16 (19)	25.64 (10)	15.79 (3)	33.33 (3)
Making Visits to Patients' Homes	0.00 (0)	4.76 (3)	20.51 (8)	15.79 (3)	22.22 (2)
Using Telemedicine Technologies	0.00 (0)	6.35 (4)	23.08 (9)	42.11 (8)	11.11 (1)

Serving as a Medical Home	5.68 (5)	53.97 (34)	28.21 (11)	15.79 (3)	33.33 (3)
[1]Column percentages are reported.					

All eighty-five providers who reported that their practice utilized a team approach to care were asked to indicate who else participated in patient care teams, excluding themselves. Provider responses are contained in Table 4c. More than half of providers reported that teams were composed of clinical geneticists, dietitians/nutritionists, social workers, nurse, and genetic counselors. Patient advocates, office/administrative managers, primary care providers, metabolic laboratory directors, and other providers were included less frequently. 'Other' providers were specified by respondents and included nurse practitioners (n=6), medical assistants (n=1), psychologists (n=1), physician assistants (n=1), and biochemical genetics fellows (n=1).

Table 4c. Team Membership (Excluding Respondent) Among Those Who Reported Working On a Team	
	Total (n=85)
Clinical Geneticist	54.12 (46)
Dietitian/Nutritionist	84.71 (72)
Social Worker	51.76 (44)
Nurse	64.71 (55)
Patient Advocate	3.53 (3)
Office/Administrative Manager	47.06 (40)
Primary Care Providers	43.53 (37)
Genetic Counselor	63.53 (54)
(Metabolic) Laboratory Director	40.00 (34)
Other	10.59 (9)

Given the importance that providers gave them in the survey, it is unsurprising that a number of comments in the Best Practices and Wrap-Up sections of the survey discussed the team approach. Six providers mentioned the importance of the multidisciplinary team approach; three simply stated:

> "The multidisciplinary approach to care helps meet the complex needs of patients and families and divides the workload."
>
> "Our team approach is very valuable to us."
>
> "When the parents of children have easy access to the coordinator, dietician, social worker, clinic staff, and the physician, everything works much better. This is especially important for children on special diets…"

In fact, three others noted other team members (a social worker, dietitian, and administration/billing person) and two others discussed the need to expand the team. One stated that the clinic has had difficulty utilizing the team approach because of the "lack of care providers interested in caring for [inborn errors of metabolism]." Two providers pointed to the importance of other team members for easing clinical geneticists' workload and as a way of addressing the shortage of clinical geneticists:

"A knowledgeable and dedicated metabolic dietician and nurse practitioner have greatly eased the load from our metabolic physicians."

"Metabolic/Genetic Nurse Practitioner/Dietitian partnerships work well for chronic management of most [inborn errors of metabolism] and should be cultivated given the increase [in] patient load and fewer biochemically trained physicians."

Of the thirty-nine providers who reported that their practice utilized satellite clinics, only twenty-seven providers responded to follow-up questions regarding these clinics. The reports of these twenty-seven providers are contained in Table 4d. Providers reported working at a variety of satellite clinics; thirty-seven reported working at two clinics, while 30% reported working at three or more clinics. However, most providers (74%) reported only working at satellite clinics one to two days per month. Providers were also asked to identify the top three reasons they practice at satellite clinics. Nearly all (96%) reported that satellite clinics were geographically easier for patients, while twenty-six percent reported that they practiced at satellite clinics because of uninsured patients.

Table 4d. Characteristics of Satellite Clinic Practice Among Those Providing Care at these Places

	Total (n=27)
Number of Satellite Clinics Practiced At	
One	33.33 (9)
Two	37.04 (10)
Three or More	29.63 (8)
Number of Days Practicing at Satellite Clinics per Month	
1-2 days	74.07 (20)
3-5 days	7.41 (2)
6-10 days	11.11 (3)
11-20 days	-
More than 20 days	3.70 (1)
Not Reported	3.70 (1)
Top Reasons for Practicing at Satellite Clinics	
Geographically Easier for Patients	96.30 (26)
Satellite Clinic Specialization	11.11 (3)
Provider Convenience	7.41 (2)
Contractual Obligations	22.22 (6)
Financial Incentives of State Mandate	18.52 (5)
Uninsured Patients	25.93 (7)
Clinics Affiliate with my Employer (e.g., outreach facility)	22.22 (6)
Personal Beliefs	14.81 (4)
Other Patient Barriers	7.41 (2)
Other	3.70 (1)

Sixteen providers reported making visits to patients' homes; however, only twelve providers answered follow-up questions regarding these visits. Their responses are contained in Table 4e. Most providers (67%) reported making home visits one to two days per month. Providers were also asked the top three reasons they made home visits. Half of providers reported that they made home visits because they were geographically easier for patients and because of other patient barriers. Those other patients barrier included a lack of transportation (n=3), a "need to understand the environment" (n=1), and to "see why [families] can't be compliant" (n=1).

Table 4e. Characteristics of Home Visit Practices Among Those Making Home Visits	
	Total (n=12)
Number of Days Making Home Visits per Month	
1-2 days	66.67 (8)
3-5 days	16.67 (2)
Not Reported	16.67 (2)
Top Reasons for Making Home Visits	
Geographically Easier for Patients	50.00 (6)
Provider Convenience	-
Contractual Obligations	-
Financial Incentives of State Mandate	8.33 (1)
Uninsured Patients	-
Personal Beliefs	25.00 (3)
Other Patient Barriers	50.00 (6)
Other	16.67 (2)

All twenty-two providers who reported that their practice utilized telemedicine responded to follow-up questions regarding telemedicine. Their responses are contained in Tables 4f and 4g. As shown in Table 4f, eighty-two percent of providers utilize remote patient monitoring and fifty-nine percent report utilizing remote patient visits Providers most frequently reported utilizing these types of telemedicine one to two days per month (44% and 46% respectively), as shown in Table 4g. Thirty-six percent of providers reported using some other type of telemedicine. Finally, half of providers who use telemedicine reported that it could be improved to assist in the care of metabolic patients. Three providers mentioned difficulty accessing telemedicine and four reported that they needed the technology and equipment to be both better and simpler. Providers also mentioned that families' comfort with telemedicine was an issue (n=1) and that telemedicine needed to be better reimbursed (n=1). Finally, two providers offered ideas of how they would like to use telemedicine technologies but currently cannot:

> "I respond to patient questions. Would be good to be more proactive in reaching them but there is insufficient time to do so."

> "It would be better if we could actually have local providers help us with the physical exam."

Table 4f. Characteristics of Telemedicine Practice among Those Who Use Telemedicine	
	Total (n=22)
Remote Patient Monitoring	
No	18.18 (4)
Yes	81.82 (18)
Remote Patient Visits	
No	40.91 (9)
Yes	59.09 (13)
Other Types of Telemedicine	
No	63.64 (14)
Yes	36.36 (8)
Reasons for Using Telemedicine	
Geographically Easier for Patients	81.82 (18)
Provider Convenience	54.55 (12)
Contractual Obligations	4.55 (1)
Financial Incentives of State Mandate	-
Uninsured Patients	-
Provider-to-Provider Communication about Patient	22.73 (5)
Personal Beliefs	9.09 (2)
Other Patient Barriers	9.09 (2)
Other	4.55 (1)
Could Telemedicine Be Improved	
No	31.82 (7)
Yes	50.00 (11)
Not Reported	18.18 (4)

Table 4g. Number of Days Per Month Providers Use Telemedicine		
	Remote Patient Monitoring (n=18)	Remote Patient Visits (n=13)
1-2 days	44.44 (8)	46.15 (6)
3-5 days	27.78 (5)	23.08 (3)
6-10 days	11.11 (2)	7.69 (1)
11-20 days	5.56 (1)	7.69 (1)
More than 20 days	5.56 (1)	-
Not Reported	5.56 (1)	15.38 (2)

Of the fifty-six providers who reported that their practice serves as a medical home, fifty-five providers responded to follow-up questions. Sixty percent of providers stated that their practice served as the medical home for patients with metabolic conditions diagnosed following a positive newborn screening. Of the twenty-two providers who stated that their practice did not serve as a medical home or were not sure if their practice served as a medical home, fifty-nine percent stated that one of the patients' other providers served as the medical home. Of those thirteen providers, twelve responded that the patients' primary care providers served as the medical home; one provider declined to answer.

5. Challenges to Providing Care

Providers were asked to respond to a series of statements regarding challenges they face providing care to patients with metabolic conditions diagnosed following a positive NBS during the first year of life. Statements reflected a variety of potential challenges, including geographic distance, clinic staffing, and educational needs and responsibilities. Response categories ranged from 'very challenging' to 'not at all challenging.' A total of 84 providers responded to at least one statement; their responses are contained in Figure 5 (Appendix C, Table 5). These analyses were not bifurcated by provider type.

Providers reported that factors associated with families were challenging. More than half of respondents reported that parental/familial knowledge and awareness of the metabolic disorder (64%) and compliance with treatment and management protocols (55%), and language barriers (52%) between providers and patients/families were somewhat challenging. Fifty-one percent of providers stated that the geographic distance between providers and families was somewhat challenging; an additional 43% stated that distance was very challenging. Three comments were made reflecting this challenge:

> "Our clinic services the entire state so we face challenges in actually getting families to clinic. Some of our patients drive from 3 or more hours away to be seen and when a condition calls for that perhaps more than every couple of months."

> "The size of the catchment area and ability to get the services to families far away."

> "Solo practice with too large a geographic area to know all the community resources as well as needed for optimal care."

Factors associated with the health care system were also considered challenging. Fifty-two percent of providers reported that having to take on multiple roles when providing care was somewhat challenging; an additional thirteen percent of providers reported it was very challenging. Primary care providers knowledge of metabolic disorders was also reported as challenging – twenty-five percent of providers reported it was very challenging and forty-eight percent of providers reported it was somewhat challenging. One provider commented that, "our attempts to educate [primary care providers] have also met with resistance." Additionally, providers frequently reported that having enough time to provide patients/families with all necessary resources was very (41%) or somewhat (44%) challenging. Forty-six percent of providers rated the ability to transition patients to hospital providers when necessary as not very challenging.

Providers reported that the most challenging aspect of their work was the lack of reimbursement for all of the care that patients and families need. Sixty-two percent of providers reported that reimbursement was very challenging. An additional 27% rated reimbursement as somewhat challenging. That providers' find reimbursement challenging was reflected in their comments at the end of the survey; ten providers specifically mentioned reimbursement. One provider summed the problem with reimbursement succinctly: "Very poor reimbursements for the clinic visits. No reimbursement for all the time spent on patient care outside of clinic. We lose money on every patient we see. We are dependent on philanthropy and the small amount the State provides to survive." Two other providers actually reported that reimbursement was not an issue because they were state employees; one stated, "I am fortunate to work in a state with a coordinated program -- we work closely with the NBS program and have funding to support [a registered dietitian, social worker], and other staff to provide comprehensive care."

Fifty-four percent of providers reported the adequacy of clinic staffing was very or somewhat challenging. Nine providers specifically mentioned staff adequacy as an issue they faced; three

mentioned general staffing issues, while three mentioned the specific lack of a physician, a social work, and a staff member in charge of billing/referral, respectively. Three providers mentioned the need for clinical geneticists trained in metabolic conditions:

> "The greatest challenge is the number of patients and the relative lack of enough qualified physicians who are able to provide knowledgeable care. We need more geneticists trained in metabolic diseases."

> "Clinical Geneticists [are] untrained in metabolism or nutrition."

> "Knowledgeable faculty to diagnose and treat rare disorders. Maybe the federal government needs to offer funding for metabolic specialists. How about mandating a metabolic specialist for every state?"

Unique insurance protocols and the availability of prescription medications and medical foods were related challenges. Seventy-seven percent of providers reported that insurance protocols were somewhat or very challenging. Eight providers mentioned general insurance issues in their comments. Providers stated:

> "Largest amount of time is spent chasing insurance issues around and that is the one area where we have no support so providers and limited office staff end up spending their time doing it."

> "Insurance issues are overwhelming at times."

> "We are all feeling overworked and underappreciated. We seem to always have to battle for our patients and their care needs as we deal in rare disorders and insurers, etc. have not heard of most of them."

Eighty-seven percent of providers reported that the availability of prescription medications and/or medical foods was somewhat or very challenging. Fourteen providers commented that working with insurers for medical foods and formulas was difficult:

> "The paperwork alone required for the medications/formulas/foods is quite time consuming."

> "Making insurers aware of the needs of metabolic [patients] specifically the need to cover medications and medical foods."

> "Fighting with insurance companies to cover for medical nutrition products."

> "Too much insurance resistance to cover formula, meds, food, testing, etc."

> "Refusal of coverage by health plan providers for medical foods, modified low protein foods, medications used to treat IEM."

> "Getting the appropriate medical beverage/ foods covered to ensure appropriate standard of care."

Not only is dealing with insurance companies to cover medical foods and formulas time consuming, but patients' quality of care and health is at stake if providers are unable to demonstrate the importance of such treatments to insurance companies.

The interconnectedness of the issues discussed above – the time spent with patients, compliance, staffing adequacy, and reimbursement – are reflected in two provider comments. These providers stated:

> "The greatest challenge is donating the necessary time to the patients. Ongoing communication and a strong bond between doctor and patient/parent is the key to successful treatment, and metabolic geneticists are in short supply but high demand."

> "Since care for these disorders is not well reimbursed, we are not well staffed. Staffing and time are constant problems."

Thirty-eight percent rated their own educational needs regarding metabolic disorders as not very challenging, despite the fact that thirty-four percent of providers reported needing periodic and twenty-seven percent reported appreciating frequent updates about metabolic disorders and care practices. One reason why providers own educational needs may not be a substantially challenging issue can be traced to the comments providers included in the survey. Four separate providers stated that they have found sharing ideas with other providers and clinics was valuable. Three of these providers specifically cited electronic methods of communication; two stated:

> "ListServes among metabolic providers has been VERY helpful for sharing information and case reports."

> "ListServes have been very VALUABLE as a way to communicate with other metabolic providers."

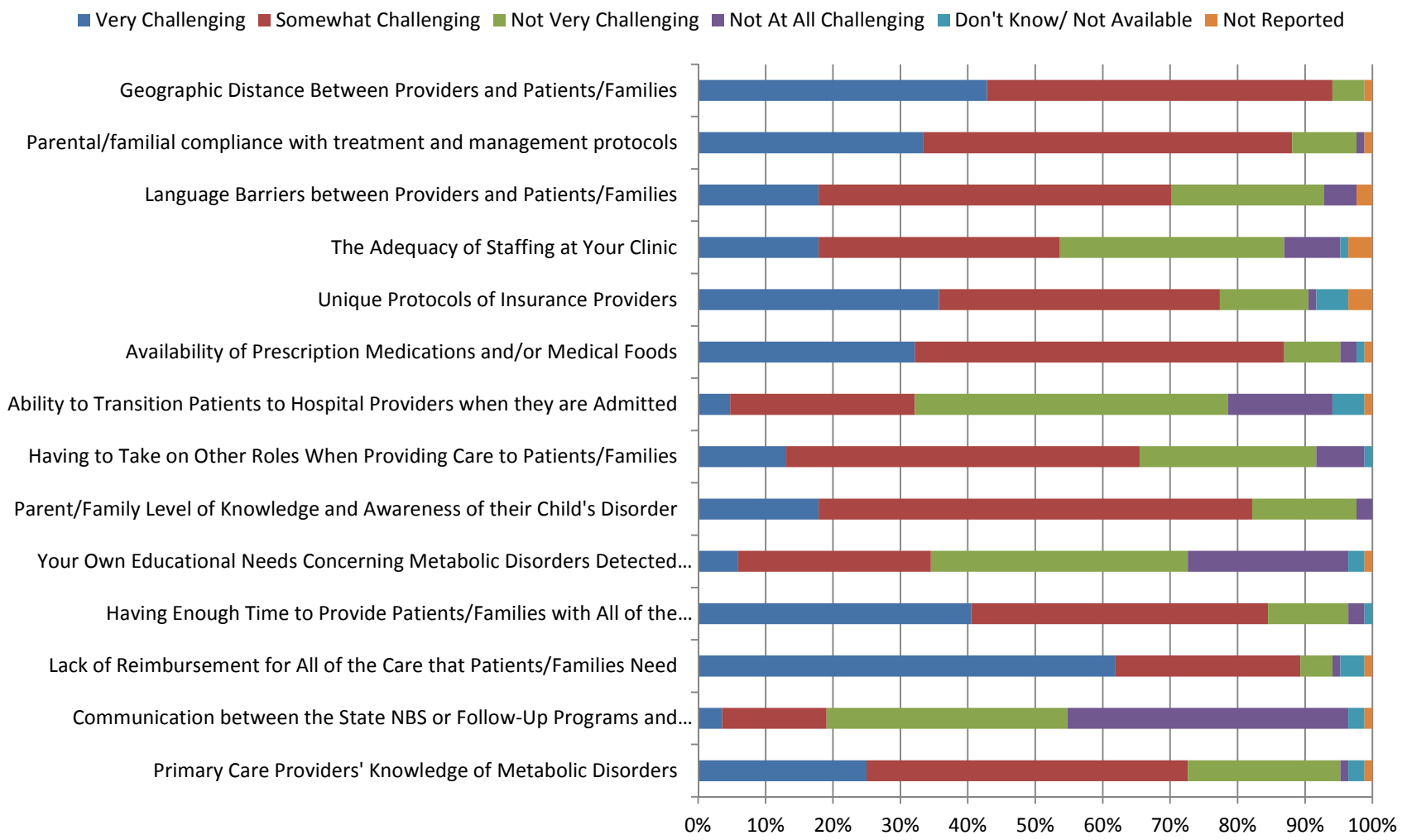
Forty-two percent of providers reported that their communication with state NBS or follow-up programs was not at all challenging. One provider stated that "a close partnership between the newborn screening program and the metabolic clinicians helps facilitate rapid interpretation of results and proper emergent management when necessary." One of the four providers who was experiencing difficulty working with NBS systems reported that "the lack of communication from the state NBS lab to our in-house NBS coordinator to the MD/RD involved in the care of metabolic patients has greatly increased over the last year [has resulted in a] delay in working with positive cases."

One other challenge was mentioned in provider comments that was not included in the survey. Four providers mentioned the lack of knowledge of the history of the disorder and lack of guidelines for care. Two providers stated:

> "A consensus on one way to treat the particular disorder. Every center treats patients differently."

> "Lack of knowledge about the prognosis and best treatments as well as the natural history of many of the disorders and the fact that there is not a systematic way to address this problem."

Figure 5. Challenges to Providing Care During the First Year of Life (n=84)



6. Education

Providers were asked to share information on the educational practices and resources they use when working with families. A total of 86 providers answered at least one question in the education section of the survey. These analyses were not examined by provider type.

First, providers were asked to indicate the three things about which they educate families. Two topics were reported by nearly all providers: treatment and management for the disorders (95%) and basic information about the disorder (90%). Approximately half of providers also reported educating families about the complications (50%) and prognosis (48%) of the disorder.

Next providers were asked about the average amount of time per month that they spend educating families of children with metabolic disorders from birth to age one. Providers most frequently reported spending between one and two hours each month (31%) educating families. Approximately 38% of providers reported spending between three and ten hours educating families each month. Seven percent of providers reported spending more than twenty hours each month educating families.

Providers were also asked to indicate the three educational materials they use most frequently with families. The majority of providers reported using pamphlets and resource binders (87%), websites (86%), and support groups (67%) to educate families. Fewer providers reported educating families with parent advocates (13%) and other materials (12%).

In their comments, two providers reported needing additional educational materials. One reported the need for easy to read educational materials for families; another discussed the need for educational materials explaining the difference between true and false positives, and borderline results. Three providers indicated that they needed more time to educate families on metabolic disorders; these providers stated:

> "…time to educate our lower literacy families on the disorders."

> "Because we have a regional program and a very small team, we don't see the children often and I think this impairs our ability to teach the families important aspects of the disorders."

> "Having the time to educate the families and primary care providers about the metabolic disorder and their role in management. "

Providers also reported that the most common resources of information that families used were websites (100%) and pamphlets (58%). Previous experience with metabolic disorders (35%) and contact with follow-up coordinators (29%) and primary care providers (22%) were also frequently cited sources of education used by families. Given the sources of education used by families, providers reported that the information families have about metabolic disorders when they visit the providers' practice is only accurate sometimes (69%).

Table 6. Educational Practices and Resources Used by Providers

	Total (n=86)
Things Providers Educate Families About	
Basic Information About the Disorder	89.53 (77)
Treatment/Management for the Disorder	95.35 (82)
Complications of the Disorder	50.00 (43)

Prognosis of the Disorder	47.67 (41)
Other Services that the Patient May Need	16.28 (14)
Insurance Protocols Pertaining to the Disorder	6.98 (6)
Other	2.33 (2)
Number of Hours Spent Educating Families per month	
Less than 1 hour	10.47 (9)
1-2 hours	31.40 (27)
3-5 hours	19.77 (17)
6-10 hours	18.60 (16)
11-20 hours	12.79 (11)
More than 20 hours	6.98 (6)
Educational Materials Providers Use Most Frequently (select three)	
Pamphlets/Resource Binders	87.21 (75)
Parent Advocates	12.79 (11)
Support Groups	67.44 (58)
Websites	86.05 (74)
Other	11.63 (10)
Most common Sources of Information Used by Families (select three)	
Websites	100.00 (86)
Previous Experience with Disorder	34.88 (30)
Pamphlets	58.14 (50)
NBS Follow-up Coordinator	29.07 (25)
Primary Care Providers	22.09 (19)
Other	12.79 (11)
How Often Information Families have about Disorders are Accurate	
Always	0.00 (0)
Often	9.30 (8)
Sometimes	68.60 (59)
Rarely	18.60 (16)
Never	0.00 (0)
Not Applicable	2.33 (2)
Not Reported	1.16 (1)

7. Relationships with Primary Care Providers

Providers were asked about the relationships between primary care providers and metabolic professionals. A total of 84 metabolic providers answered questions regarding their relationships with primary care providers; those results are presented here. Only one primary care provider was eligible for the final sample of providers used in this analysis; therefore, those results are not presented.

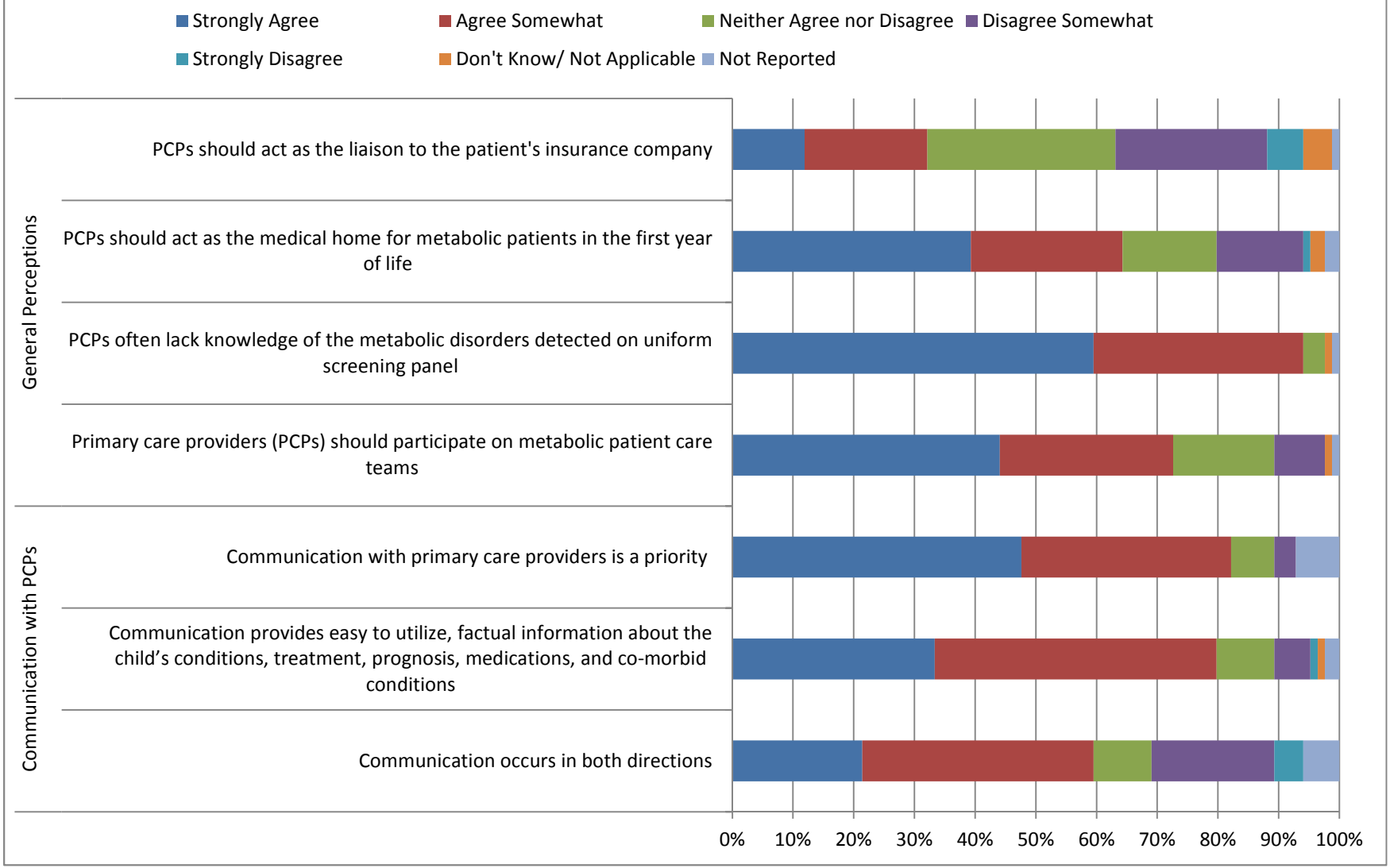
Seventy-six percent of metabolic providers reported working with primary care providers; nineteen percent reported that they did not work with primary care providers (data not shown). All metabolic providers – whether or not they worked with primary care providers – were eligible to answer

questions about their perceptions of primary care providers' knowledge and communication with primary care providers.

Metabolic providers were asked to rate their level of agreement from 'strongly agree' to 'strongly disagree' on four general perceptions about primary care providers. These are shown in Figure 7a (Appendix C, Table 7a). The majority of metabolic providers agreed or strongly agreed that providers often lack knowledge about metabolic disorders on the NBS panel (94%). Despite this, most providers also agreed or strongly agreed that primary care providers should participate on metabolic care teams (73%) and act as the medical home for patients (64%). However, only thirty-two percent of providers agreed or strongly agreed that primary care providers should act as a liaison to the insurance company for patients with metabolic disorders.

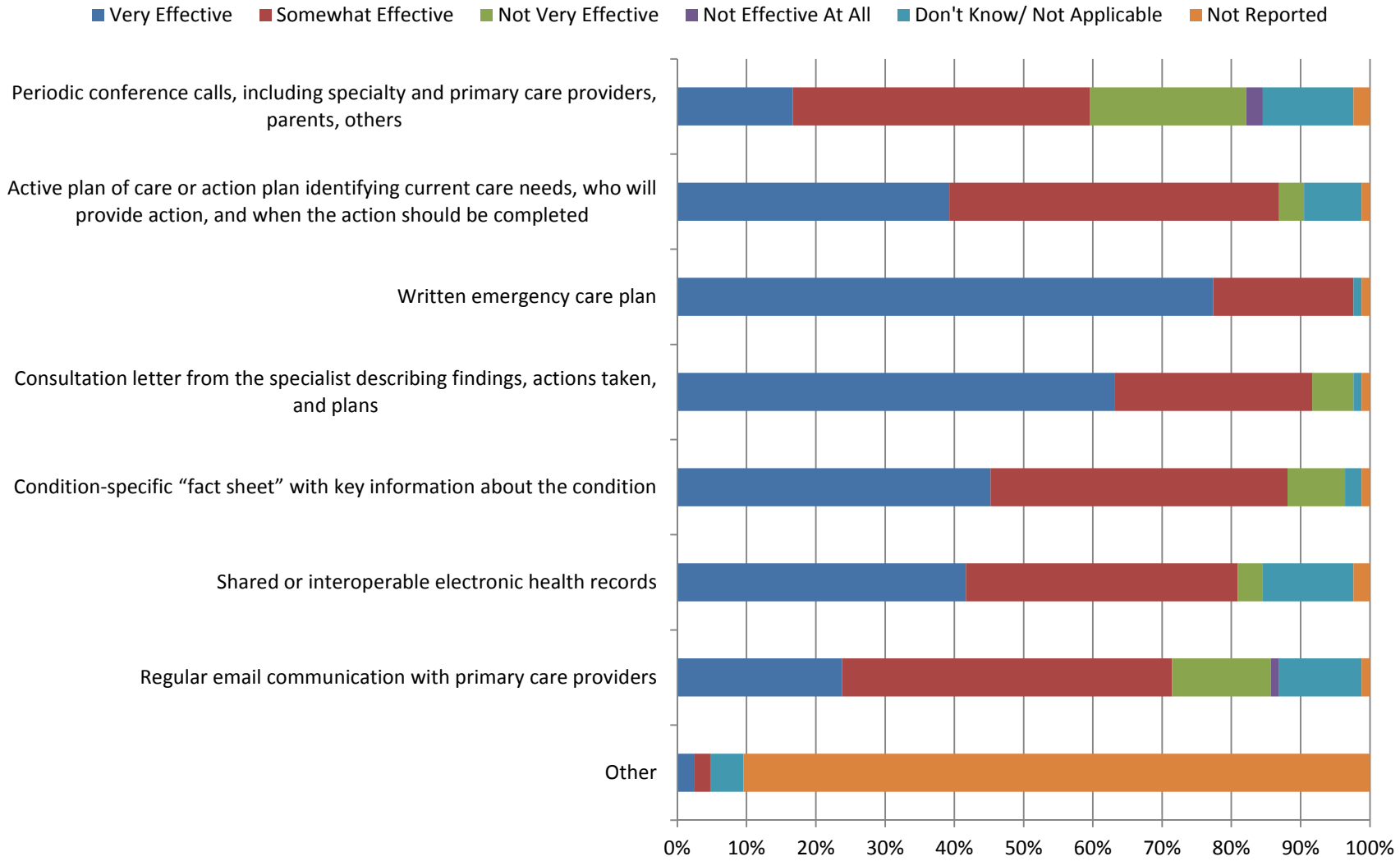
Metabolic providers were also asked to rate their level of agreement from 'strongly agree' to 'strongly disagree' on three measures of communication with primary care providers. Eighty-two percent of metabolic providers agreed or strongly agreed that communicating with primary care providers was a priority, including forty-eight percent who reported they strongly agreed. Eighty percent of metabolic providers agreed or strongly agreed that communication with primary care providers provides easy to use, factual information about the patients' health. Sixty percent agreed or strongly agreed that communication occurs both from metabolic providers to primary care providers and from primary care providers to metabolic providers.

Figure 7a. Metabolic Perceptions of Primary Care Providers Participation in Patient Care (n=84)



Metabolic providers were also asked to rate the effectiveness of methods that facilitate the sharing of patient management with primary care providers from 'very effective' to 'not effective at all.' These results are shown in Figure 7b (Appendix C, Table 7b). Metabolic providers most frequently reported written emergency care plans (77%) and consultation letters from specialists (63%) as very effective. Condition-specific fact sheets (88%) and shared or interoperable electronic health records (81%) were rated very to somewhat effective. Metabolic providers rated active care plans (48%), regular email communication (48%), and periodic conference calls (43%) most frequently as somewhat effective.

Figure 7b. Effectiveness of Methods to Share Patient Management with Primary Care Providers (n=84)



Finally, providers were asked to rank the top three ways they communicated with primary care providers after a patient's routine visit. Some providers rated more than one activity in the top three; all of these reports were taken into account in order to describe the variety of methods providers use to communicate with primary care providers. These responses are contained in Table 7c. The most common primary ways of communicating with primary care providers were phone call (41%) and fax (35%).

Table 7. Most Common Methods of Communications with PCPs (n=84)[1]

	First	Second	Third
Email	3.37 (3)	24.32 (18)	32.81 (21)
Fax	34.83 (31)	20.27 (15)	23.44 (15)
Phone Call	40.45 (36)	31.08 (23)	20.31 (13)
Postal Mail	17.98 (16)	21.62 (16)	23.44 (15)
Other	3.37 (3)	2.70 (2)	0.00 (0)

[1]Column percentages are shown.

8. Attitudes About Expanding the Newborn Screening Panel

In an effort to learn about providers attitudes about expanding the NBS panel, providers were asked to rate their level of agreement with a series of statements about the impact of expanding the panel from 'strongly agree' to 'strongly disagree.' A total of 83 providers had valid responses to at least one of these statements and were included in these analyses. Analyses are contained in Figure 8 (Appendix C, Table 8).

Providers most frequently strongly agreed that there are not currently enough providers educated to care for patients with metabolic disorders on an expanded NBS panel (63%). Additionally, metabolic providers strongly agreed that expanding the NBS panel would increase their case load (60%) and the amount of time spent treating patients (54%). Finally, approximately 40% of providers strongly agreed that they were concerned about being reimbursed for care they would need to provide to new patients (41%), and that there would not be evidence-based treatment protocols for the new conditions (41%). Sixty-four percent of providers were also concerned that an expanded NBS panel would result in more false-positive disorders being detected. Five providers reported specific concerns about expanding the NBS panel due to a lack of treatment protocols and advanced testing. They stated:

> "There ought to be nationalization of standards of NBS as well as the creation of a budget to support the effort. The model to follow might be the national highway system."

> "Having treatment guidelines, when they are available, is helpful but I understand that there isn't always enough evidence to develop a definitive protocol."

> "This will all become very ugly and people will have even less trust in the medical establishment. Adding Comparative Genomic Hybridization will be a disaster. We need more research on what these variants of unclear significance mean."

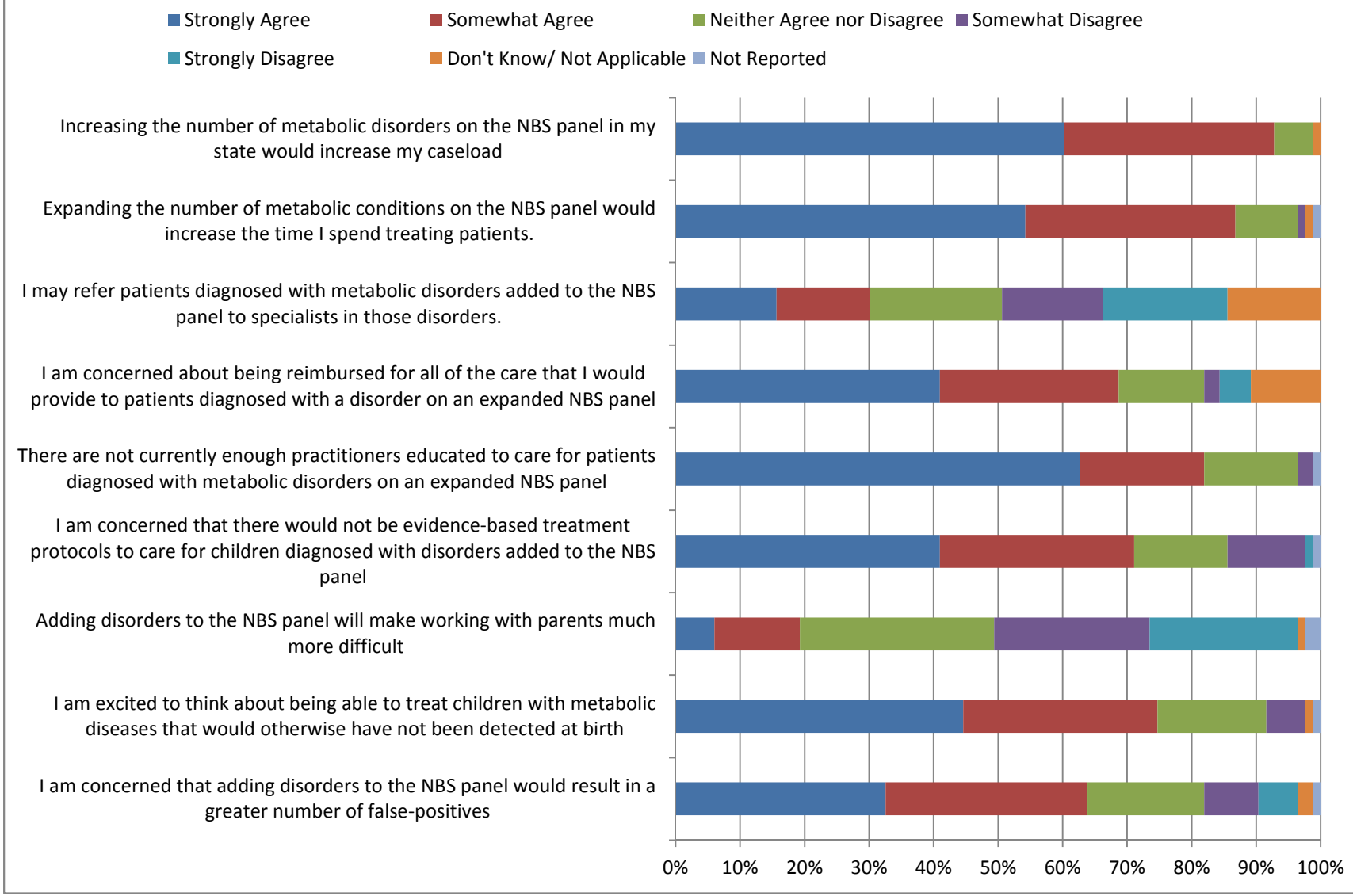
"A lack of evidence-based treatment strategies. This is also my biggest concern with regards to expanding the number of disorders on the screening panel. I am not in favor of identifying disorders for which there is no effective therapy."

"Lack of knowledge on the significance, natural history and treatment of many of the expanded disorders, "

Despite these concerns, seventy-five percent of providers were excited about treating children with metabolic disorders who would otherwise not have received treatment. Providers also did not report being concerned that an expanded would make working with parents more difficult; forty-seven percent disagreed or strongly disagreed with the statement and an additional thirty percent reported that they neither agreed nor disagreed.

One statement garnered reports that were relatively spread. In response to the statement "I may refer patients diagnosed with metabolic disorders added to the NBS panel to specialists in those disorders", thirty percent of providers agreed or strongly agreed, thirty-five percent disagreed or strongly disagreed and twenty-one percent neither agreed nor disagreed.

Figure 8. Attitudes Toward Expanding the NBS Panel (n=83)



Next Steps and limitations:

This survey was conducted on a broad spectrum of providers who treat patients with metabolic disorders, but it is not generalizable to all metabolic providers due to selective response. Nonetheless, the responses here mirror strongly those found in the first phase interview pilot project.

Two themes emerge here. One is that the extra-care process post NBS, specifically care coordination and education, needs to be investigated for effectiveness and best practices. Care management and payment management need to be the core functions of at least one team member. The field also needs to find more consistent and effective ways of educating families and primary care providers.

Secondly is that workforce issues need to be addressed. The current consensus is that resources are inadequate to handle the intensity of care volume as it currently exists. Expansions to the metabolic NBS panel without first addressing the ineffectiveness of the current models of care will only exacerbate these issues. Explored should be reconfigurations to the care teams, better care protocols, and standard methods of communication across the spectrum of care. The use of technology and integration into other best practice methods should be a priority.

Appendix A. Survey

UNIVERSITY of NEW HAMPSHIRE

A National Assessment of the Newborn Screening Workforce

Informed Consent Information

You have been invited to participate in a research project that will examine the process of care for patients diagnosed with metabolic conditions that are detected on newborn screen (NBS) from diagnosis to the first birthday. While you will not receive any direct benefits from your participation, the information gathered here will be valuable for informing the current and ongoing policy discussions regarding reimbursement for genetic services.

This project is being conducted by Robert J. McGrath, Associate Professor in the Department of Health Management and Policy at the University of New Hampshire (UNH). This project is funded by the National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives (NCC). This survey asks you questions regarding the process of care and challenges to providing care for average patients with metabolic conditions. You will not be asked to describe any specific cases. You will also be asked about how you believe an expanded NBS panel will impact your work.

Your participation is voluntary, and you may withdraw your consent and discontinue participation at any time. Although we hope that you will answer every question, you are certainly free to skip any questions. Your responses will be kept confidential. The survey data will only be analyzed and reported in aggregated ways that will not reveal your identity. Thus, no individual identity will be determinable through demographic variables such as zip code. The results may be used in reports, publications, and/or presentations.

This project will be successful only if you help us. We know that you are extremely busy, but please take 20 minutes to complete the confidential survey. Because of the highly specialized field of caregivers we are interviewing, you are one of approximately 200 health care providers who will be asked to participate in this research. Participation in this study is expected to present minimal risk to you.

If at any time you have questions or concerns about any procedure in this project, you may e-mail the investigator (Robert.McGrath@unh.edu) or speak with the investigator by calling 603-862-5047. You should also understand that you will be able to request a summary of the findings. If you have questions about your rights as a research subject, you may contact Julie Simpson in UNH Research Integrity Services, by phone 603-862-2003 or email julie.simpson@unh.edu.

Instructions

Thank you for participating in this survey of genetic service providers who treat children with metabolic disorders. This survey is being conducted by Robert J. McGrath, Associate Professor of Health Management and Policy at the University of New Hampshire, and is funded by the National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives.

This survey seeks to understand the scope and intensity of services provided to patients with metabolic disorders and their families by all caregivers from the time of positive newborn screen (NBS) and subsequent diagnosis of a metabolic disorder, through the first year of life.

We take a broad view of both the care provided, including time spent with the patient and time spent working on the patient's behalf, and the responsibilities of people providing care, including the work of clinical providers and office staff. We are also interested in your views on the potential consequences of adding tests to your state's current NBS panel. Your input is highly valuable in informing current policy discussions nationally.

Thank you for your time and participation.

1. What is your primary professional role?

- ❑ Clinical Geneticist
- ❑ Dietitian/Nutritionist
- ❑ Social Worker
- ❑ Nurse
- ❑ Other (specify) _____
- ❑ Patient Advocate **[SKIP TO Section 2, Page 8, question 36]**
- ❑ Office/Administrative Manager **[SKIP TO Part II, Page 6, question 25]**
- ❑ Genetic Counselor
- ❑ Primary Care Provider

2. **Approximately what percentage of your practice/work is dedicated to treating children who have been identified through NBS and subsequently confirmed to have a metabolic disorder?**

- ❑ 0-10%
- ❑ 11-20%
- ❑ 21-30%
- ❑ 31-40%
- ❑ 41-50%
- ❑ 51-60%
- ❑ 61-70%
- ❑ 71-80%
- ❑ 81-90%
- ❑ 91-100%

Section 1: Process of Care

In order to assess the scope and intensity of services that you provide to patients with metabolic disorders, we distinguish among 3 categories of metabolic disorders: organic acid disorders, fatty acid oxidation disorders, and amino acid disorders. We recognize that there is substantial variation within each category of disorder; however, please answer the following questions with regards to an average patient in your practice diagnosed with each type of reference metabolic disorder after a positive NBS from birth through the first year of life.

Also, for all questions, please describe only YOUR workload and not the workload of other professionals with whom you may work.

While we recognize that not every state tests for all of the disorders on the Uniform NBS panel, we ask only about the disorders on the panel in order to gain a broad understanding of processes of care across the United States.

Part I. Processes of Care by Metabolic Disorder Type

3. **Do you treat children diagnosed with the following categories of metabolic disorders? Please check all that apply.**

- ❑ Organic Acid Disorders (for example, Propionic Academia) **[IF CHECKED - ANSWER PART 1A, pg. 3]**
- ❑ Fatty Acid Oxidation Disorders (for example, Carnitine Uptake Defect / Carnitine Transport Defect) **[IF CHECKED - ANSWER PART 1B, pg. 4]**
- ❑ Amino Acid Disorders (for example, Classic Phenylketonuria) **[IF CHECKED - ANSWER PART 1C, pg. 5]**
- ❑ I do not treat children diagnosed with any of these types of disorders. **[IF CHECKED - SKIP TO PART II, pg. 6]**

Part IA. Processes of Care for Organic Acid Disorders

For the following questions, please think of an average patient in your practice who has been diagnosed with a disorder of organic academia from the time of positive screen through the first year of life.

4. **Please indicate up to 3 disorders of organic academia that you see most often in your practice.**

- ❑ Propionic Academia (PROP)
- ❑ Methylmalonic Acidemia (Methylmalonyl-CoA Mutase) (MUT)
- ❑ Methylmalonic Acidemia (Cobalamin Disorders) (Cbl, A, B)
- ❑ Isovaleric Acidemia (IVA)
- ❑ 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)
- ❑ 3-Hydroxy-3-Methyglutaric Aciduria (HMG)
- ❑ Holocarboxylase Synthase Deficiency (MCD)
- ❑ B-Ketothiolase Deficiency (BKT)
- ❑ Glutaric Acidemia Type I (GA1)
- ❑ Not Applicable

5. **How soon after the NBS lab detects an abnormal blood result do you typically talk to a patient's family on the phone for the first time?**

- ❑ The Same or Next Day
- ❑ Within the Week
- ❑ Within Two Weeks
- ❑ Within the Month
- ❑ Within Six Months
- ❑ Later than Six Months

6. **Approximately how long is your first phone call with the patient's family?**

- ❑ Less than 1 Hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ 4-6 Hours
- ❑ More than 6 Hours

7. **How soon after the NBS lab detects an abnormal blood result do you typically see a patient and their family for the first time?**

- ❑ The Same or Next Day
- ❑ Within the Week
- ❑ Within Two Weeks
- ❑ Within the Month
- ❑ Within Six Months
- ❑ Later than Six Months

8. **Approximately how long is your first visit with a patient and their family?**

- ❑ Less than 1 Hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ 4-6 Hours
- ❑ More than 6 Hours

9. **How frequently do you see a patient and their family for follow-up after the first visit during the…**

	More than Once a Week	***Once a Week***	***2-3 Times per Month***	***Once a Month***	***Every Couple of Months***	***Never***
First Month	❑	❑	❑	❑	❑	❑
Months 2-3	❑	❑	❑	❑	❑	❑
Months 4-6	❑	❑	❑	❑	❑	❑
Months 7-12	❑	❑	❑	❑	❑	❑

10. **On average, how long are follow-up visits with you during the first year of life?**

- ❑ Less than 15 Minutes
- ❑ 15-30 Minutes
- ❑ 30-45 Minutes
- ❑ 45 Minutes-1 Hour
- ❑ More than 1 Hour

Part IB. Processes of Care for Fatty Acid Oxidation Disorders

For the following questions, please think of an average patient in your practice who has been diagnosed with a disorder of fatty acid oxidation from the time of positive screen through the first year of life.

11. **Please indicate up to 3 disorders of fatty acid oxidation that you see most often in your practice.**
- ❑ Carnitine Uptake Defect/Carnitine Transport Defect (CUD)
- ❑ Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
- ❑ Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
- ❑ Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
- ❑ Trifunctional Protein Deficiency (TFP)

12. **How soon after the NBS lab detects an abnormal blood result do you typically talk to a patient's family on the phone for the first time?**
- ❑ The Same or Next Day
- ❑ Within the Week
- ❑ Within Two Weeks
- ❑ Within the Month
- ❑ Within Six Months
- ❑ Later than Six Months

13. **Approximately how long is your first phone call with the patient's family?**
- ❑ Less than 1 Hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ 4-6 Hours
- ❑ More than 6 Hours

14. **How soon after the NBS lab detects an abnormal blood result do you typically see a patient and their family for the first time?**
- ❑ The Same or Next Day
- ❑ Within the Week
- ❑ Within Two Weeks
- ❑ Within the Month
- ❑ Within Six Months
- ❑ Later than Six Months

15. **Approximately how long is your first visit with a patient and their family?**
- ❑ Less than 1 Hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ 4-6 Hours
- ❑ More than 6 Hours

16. **How frequently do you see a patient and their family for follow-up after the first visit during the…**

	More than Once a Week	***Once a Week***	***2-3 Time per Month***	***Once a Month***	***Every Couple of Months***	***Never***
First Month	❑	❑	❑	❑	❑	❑
Months 2-3	❑	❑	❑	❑	❑	❑
Months 4-6	❑	❑	❑	❑	❑	❑
Months 7-12	❑	❑	❑	❑	❑	❑

17. **On average, how long are follow-up visits with you during the first year of life?**
- ❑ Less than 15 Minutes
- ❑ 15-30 Minutes
- ❑ 30-45 Minutes
- ❑ 45 Minutes-1 Hour
- ❑ More than 1 Hour

Part IC. Processes of Care for Amino Acid Disorders

For the following questions, please think of an average patient in your practice who has been diagnosed with a disorder of amino acid metabolism from the time of positive screen through the first year of life.

18. **Please indicate up to 3 disorders of amino acid metabolism that you see most often in your practice.**
- ❑ Argininosuccinic Aciduria (ASA)
- ❑ Citrullinemia, Type I (CIT)
- ❑ Maple Syrup Urine Disease (MSUD)
- ❑ Homocystinuria (HCY)
- ❑ Classic Phenylketonuria (PKU)
- ❑ Tyrosinemia, Type I (TYR I)

19. **How soon after the NBS lab detects an abnormal blood result do you typically talk to a patient's family on the phone for the first time?**
- ❑ The Same or Next Day
- ❑ Within the Week
- ❑ Within Two Weeks
- ❑ Within the Month
- ❑ Within Six Months
- ❑ Later than Six Months

20. **Approximately how long is your first phone call with the patient's family?**
- ❑ Less than 1 Hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ 4-6 Hours
- ❑ More than 6 Hours

21. **How soon after the NBS lab detects an abnormal blood result do you typically see a patient and their family for the first time?**
- ❑ The Same or Next Day
- ❑ Within the Week
- ❑ Within Two Weeks
- ❑ Within the Month
- ❑ Within Six Months
- ❑ Later than Six Months

22. **Approximately how long is your first visit with a patient and their family?**
- ❑ Less than 1 Hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ 4-6 Hours
- ❑ More than 6 Hours

23. **How frequently do you see a patient and their family for follow-up after the first visit during the…**

	More than Once a Week	*Once a Week*	*2-3 Time per Month*	*Once a Month*	*Every Couple of Months*	*Never*
First Month	❑	❑	❑	❑	❑	❑
Months 1-3	❑	❑	❑	❑	❑	❑
Months 3-6	❑	❑	❑	❑	❑	❑
Months 6-12	❑	❑	❑	❑	❑	❑

24. **On average, how long are follow-up visits with you during the first year of life?**
- ❑ Less than 15 Minutes
- ❑ 15-30 Minutes
- ❑ 30-45 Minutes
- ❑ 45 Minutes-1 Hour
- ❑ More than 1 Hour

Part II. Non-Visit Administrative Time

For each of the following tasks, please estimate the amount of time you personally spend performing each of these non-visit activities per month during an average patient's first year of life. These questions do NOT distinguish between disorders of organic acid, fatty acid oxidation, or amino acid.

Thinking about an average patient in your practice diagnosed with a metabolic disorder detected on NBS from the time of positive screen through the first year of life...

- Dieticians ONLY – ALL others go to page 7

25. **What percentage of your patients and their families, when asked, provide a 3-day diet recall record?**

- ❑ 0-10%
- ❑ 11-20%
- ❑ 21-30%
- ❑ 31-40%
- ❑ 41-50%
- ❑ 51-60%
- ❑ 61-70%
- ❑ 71-80%
- ❑ 81-90%
- ❑ 91-100%

26. **On average, how long do you spend preparing a 3-day diet recall?**

- ❑ Less than 15 Minutes
- ❑ 15-30 Minutes
- ❑ 30-45 Minutes
- ❑ 45 Minutes-1 Hour
- ❑ More than 1 Hour

27. **What do you do when patients and their families do not provide diet recall records? Please select all that apply.**

- ❑ Complete Diet Recall with Them
- ❑ Ask Them to Complete a Diet Recall during the Visit and Review it with Them That Day
- ❑ Ask them to Complete a Diet Recall At Home and Send it to the Office for Review and Follow-up
- ❑ Other (Specify) _____

- ALL Providers and Administrative/Office Staff -

28. **About how long do you spend talking on the phone with the family per month?**

- ❑ Less than 30 Minutes
- ❑ 30 Minutes-1 hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ More than 4 Hours
- ❑ I don't do this

29. **About how long do you spend doing case management per month?**

- ❑ Less than 30 Minutes
- ❑ 30 Minutes-1 hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ More than 4 Hours
- ❑ I don't do this

30. **Do you spend time interacting with insurance companies about patients with metabolic disorders?**

- ❑ No **[SKIP TO question 33]**
- ❑ Yes

31. **About how long do you spend interacting with insurance companies per month?**

- ❑ Less than 30 Minutes
- ❑ 30 Minutes-1 hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ More than 4 Hours
- ❑ I don't do this

32. **Please rank the 3 most time-intensive reasons you interact with insurance companies?**

	First	***Second***	***Third***
General Reimbursements for Services	❑	❑	❑
Patient Eligibility for Care	❑	❑	❑
Approvals for Medical Foods/Formulas	❑	❑	❑
Approvals for Medications	❑	❑	❑
Approvals for Durable Medical Equipment	❑	❑	❑
Approval for Hospital Services	❑	❑	❑
Other _____	❑	❑	❑

33. **About how long do you spend preparing for face-to-face visits per month? Please include time that you spend meeting with medical staff, learning about the disorder, reviewing medical records, etc. but do not take into account time that you spend doing administrative tasks or working with insurance companies.**

- ❑ Less than 30 Minutes
- ❑ 30 Minutes-1 hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ More than 4 Hours
- ❑ I don't do this

34. **About how long do you spend doing other administrative tasks not already considered per month?**

- ❑ Less than 30 Minutes
- ❑ 30 Minutes-1 hour
- ❑ 1-2 Hours
- ❑ 2-4 Hours
- ❑ More than 4 Hours
- ❑ I don't do this **[SKIP TO SECTION 2, Next Page]**

35. **Please describe these other activities and the amount of time (per month) that each of these activities takes.**

_____

_____

Section 2: Care Coordination

Part I. General

Thinking about an average patient in your practice diagnosed with a metabolic disorder detected on NBS from the time of positive screen through the first year of life…

36. **What are your primary responsibilities when working with patients and their families? Please rank your top 3 responsibilities.**

	First	*Second*	*Third*
Coordinate Care for Patient and Family	❑	❑	❑
Interact with Insurance Companies for Patients	❑	❑	❑
Advocate for Patient Services	❑	❑	❑
Manage Dietary Needs	❑	❑	❑
Manage Medical Needs	❑	❑	❑
Educate Patients and Families	❑	❑	❑
Identify and/or Refer Patients to Community Resources, Support Groups, etc.	❑	❑	❑
Communicate with Primary Care Providers	❑	❑	❑
Communicate with Specialists, Hospital Staff, etc.	❑	❑	❑
Other _____	❑	❑	❑

37. **Is there someone at your clinic who works specifically as a care coordinator?**

❑ No ❑ Yes

38. **Practices/clinics use different models to meet the needs of their patients and their patients' families. Please rank the following practice models in the order of their importance to you in caring for patients with metabolic disorders and their families.**

	Most Important	*2nd Most Important*	*3rd Most Important*	*4th Most Important*	*Least Important*	*I/My practice does not do this*	
Using a Team Approach to Care	❑	❑	❑	❑	❑	❑	**SKIP TO Part II, pg. 9**
Practicing at Satellite Clinics	❑	❑	❑	❑	❑	❑	**SKIP TO Part III, pg. 9**
Making Visits to Patients' Homes	❑	❑	❑	❑	❑	❑	**SKIP TO Part IV, pg. 9**
Using Telemedicine Technologies	❑	❑	❑	❑	❑	❑	**SKIP TO Part V, pg. 10**
Serving as a Medical Home	❑	❑	❑	❑	❑	❑	**SKIP TO Part VI, pg. 11**

For each practice model that you or your practice use,
Please respond to ***EACH*** of the corresponding sections on that model.
If you use ***MORE*** than ***ONE*** Model, please answer question 53 in addition to each section.
If you do not use ***ANY*** please skip to Part VII (question 57, page 12).

Thinking about an average patient in your practice diagnosed with a metabolic disorder detected on NBS from the time of positive screen through the first year of life…

Part II. Team Approach to Care

39. **Excluding yourself, who else is part of the care team for patients diagnosed with metabolic disorders? Please check all that apply**

- ❑ Clinical Geneticist
- ❑ Dietitian/Nutritionist
- ❑ Social Worker
- ❑ Nurse
- ❑ Patient Advocate
- ❑ Office/Administrative Manager
- ❑ Primary Care Provider
- ❑ Genetic Counselor
- ❑ (Metabolic) Laboratory Director
- ❑ Other (Specify) _____

Part III. Satellite Clinics

40. **At how many satellite clinics do you treat metabolic patients?**

- ❑ One
- ❑ Two
- ❑ Three or More
- ❑ Not Applicable

41. **On average, how many days per month do you practice in satellite clinics and treat patients with metabolic disorders?**

- ❑ 1 – 2 days
- ❑ 3 – 5 days
- ❑ 6 – 10 days
- ❑ 11 – 20 days
- ❑ More than 20 days

42. **Please check the top 3 reasons you practice in satellite clinics.**

- ❑ Geographically easier for patients
- ❑ Satellite clinic specialization
- ❑ Provider convenience
- ❑ Contractual obligations
- ❑ Financial incentives of state mandate
- ❑ Uninsured patients
- ❑ Clinics affiliate with my employer (e.g. outreach facility)
- ❑ Personal belief
- ❑ Other patient barriers (specify) _____
- ❑ Other (specify) _____

Part IV. Home Visits

43. **How many days per month do you make home visits?**

- ❑ 1 – 2 days
- ❑ 3 – 5 days
- ❑ 6 – 10 days
- ❑ 11 – 20 days
- ❑ More than 20 days

44. **Please check the top 3 reasons why you make home visits.**

- ❑ Geographically easier for patients
- ❑ Provider convenience
- ❑ Contractual obligations
- ❑ Financial incentives of state mandate
- ❑ Uninsured patients
- ❑ Personal belief
- ❑ Other patient barriers (specify) _____
- ❑ Other (specify) _____

Thinking about an average patient in your practice diagnosed with a metabolic disorder detected on NBS from the time of positive screen through the first year of life…

Part V. Telemedicine

45. **Do you use remote patient monitoring, for example, remote diet recall or blood testing (i.e. heel sticks), to care for patients diagnosed with metabolic disorders?**

- ❑ No **[SKIP TO question 47]**
- ❑ Yes

46. **How many days per month do you use the remote patient monitoring?**

- ❑ 1 – 2 days
- ❑ 3 – 5 days
- ❑ 6 – 10 days
- ❑ 11 – 20 days
- ❑ More than 20 days

47. **Do you use remote patient visits, for example through internet or phone conferencing, to care for patients diagnosed with metabolic disorders?**

- ❑ No **[SKIP TO question 49]**
- ❑ Yes

48. **How many days per month do you use the remote patient visits?**

- ❑ 1 – 2 days
- ❑ 3 – 5 days
- ❑ 6 – 10 days
- ❑ 11 – 20 days
- ❑ More than 20 days

49. **Do you use other types of telemedicine (not remote patient monitoring or remote patient visits) to care for patients diagnosed with metabolic disorders?**

- ❑ No **[SKIP TO question 51]**
- ❑ Yes

50. **Please describe the other types of telemedicine that you use and how frequently you use each type per month.**

_____

_____

51. **Please check the top 3 reasons why you use telemedicine to care for patients with metabolic disorders.**

- ❑ Geographically easier for patients
- ❑ Provider convenience
- ❑ Contractual obligations
- ❑ Financial incentives of state mandate
- ❑ Uninsured patients
- ❑ Provider-to-provider communication about patient
- ❑ Personal belief
- ❑ Other patient barriers (specify) _____
- ❑ Other (specify) _____

52. **Could telemedicine be improved to assist you in the care of patients with metabolic disorders?**

- ❑ No
- ❑ Yes (specify) _____
  _____

Thinking about an average patient in your practice diagnosed with a metabolic disorder detected on NBS from the time of positive screen through the first year of life…

- ALL Providers -

53. **Thinking about the variety of practice models discussed here (satellite clinics, home visits, and telemedicine) that are used in the care of patients diagnosed with metabolic disorders, do you tend to see the same patients in only one setting or in a variety of settings?**

For example, do you utilize satellite clinic visits and telemedicine for the same patient and their family or is it more likely that you only see the patient and their family in one setting (for example, a satellite clinic) during their first year of life?

_____

_____

Part VI. Medical Home

54. **Does your practice serve as a medical home to patients with metabolic disorders?**

❑ No ❑ Yes **[SKIP TO Part VII, question 57]** ❑ Don't Know

55. **Does one of the other the patient's providers most often serve as a medical home?**

❑ No (Skip to Part VII, question 57) ❑ Yes ❑ Don't Know (Skip to Part VII, question 57)

56. **Which of the patient's other providers serves as the patient's medical home?**

- ❑ Clinical Geneticist
- ❑ Dietitian/Nutritionist
- ❑ Social Worker
- ❑ Nurse
- ❑ Patient Advocate
- ❑ Office/Administrative Manager
- ❑ Primary Care Providers
- ❑ Genetic Counselor
- ❑ (Metabolic) Laboratory Director
- ❑ Other (specify) _____

Part VII. Challenges to Providing Care during the First Year of Life

57. **Please rate how challenging you find each of the following issues when providing care to patients diagnosed with a metabolic disorder after a positive NBS and their families during the first year of life.**

	Very Challenging	*Somewhat Challenging*	*Not Very Challenging*	*Not at all Challenging*	*Don't Know / Not Available*
Geographic distance between providers and patients/families	❑	❑	❑	❑	❑
Parental/familial compliance with treatment and management protocols	❑	❑	❑	❑	❑
Language barriers between providers and patients/families	❑	❑	❑	❑	❑
The adequacy of staffing at your clinic	❑	❑	❑	❑	❑
Unique protocols of insurance providers	❑	❑	❑	❑	❑
Availability of prescription medications and/or medical foods	❑	❑	❑	❑	❑
Ability to transition patients to hospital providers when they are admitted	❑	❑	❑	❑	❑
Having to take on other roles when providing care to patients/families	❑	❑	❑	❑	❑
Parent/family level of knowledge and awareness of their child's disorder	❑	❑	❑	❑	❑
Your own educational needs concerning metabolic disorders detected on NBS	❑	❑	❑	❑	❑
Having enough time to provide patients/families with all of the resources they need	❑	❑	❑	❑	❑
Lack of reimbursement for all of the care that patients/families need	❑	❑	❑	❑	❑
Communication between the State NBS or Follow-up Programs and providers	❑	❑	❑	❑	❑
Primary care providers' knowledge of metabolic disorders	❑	❑	❑	❑	❑

Section 4: Education

Thinking about an average patient in your practice diagnosed with a metabolic disorder detected on NBS from the time of positive screen through the first year of life…

Part I. Family

58. **Please check the top 3 things that you educate families about.**

- ❑ Basic information about the disorder (e. g. natural history, genetics)
- ❑ Treatment/management for the disorder
- ❑ Complications of the disorder
- ❑ Prognosis of the disorder
- ❑ Other services that the patient may need (e. g. support groups, community resources)
- ❑ Insurance protocols pertaining to the disorder
- ❑ Other (specify) _____

59. **On average, how many hours per month do you spend educating families?**

- ❑ Less than 1 Hour
- ❑ 1 - 2 Hours
- ❑ 3 - 5 Hours
- ❑ 6 - 10 Hours
- ❑ 11 – 20 Hours
- ❑ More than 20 Hours

60. **Please select the 3 educational materials you use most frequently to teach families about their child's disorder.**

- ❑ Pamphlets/resource binders
- ❑ Parent advocates
- ❑ Support groups
- ❑ Websites
- ❑ Other (Specify) _____
- ❑ None of the above
- ❑ Not Applicable

61. **Please rank the top 3 sources of information that families most frequently use to learn about their child's metabolic disorder.**

- ❑ Websites
- ❑ Previous experience with the Disorder (for example, another family member or friend with the disorder)
- ❑ Pamphlets
- ❑ NBS follow-up coordinator
- ❑ Primary care providers
- ❑ Other (Specify) _____
- ❑ Not Applicable

62. **When you meet with families, how often would you say that the information they have about metabolic disorders when they come to your practice is accurate?**

- ❑ Always
- ❑ Often
- ❑ Sometimes
- ❑ Rarely
- ❑ Never
- ❑ Not Applicable

Part II. Relationships with Primary Care Providers

Please answer the following questions about your relationships with primary care providers with regard to the treatment of patients diagnosed with metabolic disorders after a positive NBS and their families during the first year of life.

Answer ONLY if NOT a Primary Care Provider

63. **Do you personally work with primary care providers?**

❑ No ❑ Yes

64. **What is your level of agreement with the following statements:**

	Strongly Agree	***Agree Somewhat***	***Neither Agree nor Disagree***	***Disagree Somewhat***	***Strongly Disagree***	***Don't Know / Not Applicable***
Primary care providers (PCPs) should participate on metabolic patient care teams	❑	❑	❑	❑	❑	❑
PCPs often lack knowledge of the metabolic disorders detected on uniform screening panel	❑	❑	❑	❑	❑	❑
PCPs should act as the medical home for metabolic patients in the first year of life	❑	❑	❑	❑	❑	❑
PCPs should act as the liaison to the patient's insurance company	❑	❑	❑	❑	❑	❑

65. **Please rate how effective you believe each of the following methods would be to facilitate the sharing of patient management with a primary care provider…**

	Very Effective	***Somewhat Effective***	***Not Very Effective***	***Not Effective At All***	***Don't Know / Not Applicable***
Periodic conference calls, including specialty and primary care providers, parents, others	❑	❑	❑	❑	❑
Active plan of care or action plan identifying current care needs, who will provide action, and when the action should be completed	❑	❑	❑	❑	❑
Written emergency care plan	❑	❑	❑	❑	❑
Consultation letter from the specialist describing findings, actions taken, and plans	❑	❑	❑	❑	❑
Condition-specific “fact sheet” with key information about the condition	❑	❑	❑	❑	❑
Shared or interoperable electronic health records	❑	❑	❑	❑	❑
Regular email communication with primary care providers	❑	❑	❑	❑	❑
Other (Specify)_____	❑	❑	❑	❑	❑

Please answer the following questions about your relationships with primary care providers with regard to the treatment of patients diagnosed with metabolic disorders after a positive NBS and their families during the first year of life.

66. **What is your level of agreement with the following statement about your communication with primary care providers:**

	Strongly Agree	***Agree Somewhat***	***Neither Agree nor Disagree***	***Disagree Somewhat***	***Strongly Disagree***	***Don't Know / Not Applicable***
Communication occurs in both directions	❑	❑	❑	❑	❑	❑
Communication provides easy to utilize, factual information about the child's conditions, treatment, prognosis, medications, and co-morbid conditions	❑	❑	❑	❑	❑	❑
Communication with primary care providers is a priority	❑	❑	❑	❑	❑	❑

67. **Please rank the top 3 ways you communicate with primary care providers after a patient has a routine visit?**

	1st	***2nd***	***3rd***
Email	❑	❑	❑
Fax	❑	❑	❑
Phone Call	❑	❑	❑
Postal Mail	❑	❑	❑
Other _____	❑	❑	❑

Answer ONLY *if a* Primary Care Provider

68. **What is your level of agreement with the following statement about your communication with specialty care providers:**

	Strongly Agree	***Agree Somewhat***	***Neither Agree nor Disagree***	***Disagree Somewhat***	***Strongly Disagree***	***Don't Know / Not Applicable***
Communication occurs in both directions	❑	❑	❑	❑	❑	❑
Communication does not explicitly define my role and responsibilities	❑	❑	❑	❑	❑	❑
Communication provides easy to utilize, factual information about the child's conditions, treatment, prognosis, medications, and co-morbid conditions	❑	❑	❑	❑	❑	❑
Communication provides useful information about the emergency care	❑	❑	❑	❑	❑	❑

Section 5: Attitudes about Expanding the NBS Panel

69. **Please indicate your level of agreement with the following statements regarding NBS.**

	Strongly Agree	*Somewhat Agree*	*Neither Agree nor Disagree*	*Somewhat Disagree*	*Strongly Disagree*	*Don't Know / Not Applicable*
Increasing the number of metabolic disorders on the NBS panel in my state would increase my caseload	❑	❑	❑	❑	❑	❑
Expanding the number of metabolic conditions on the NBS panel would increase the time I spend treating patients.	❑	❑	❑	❑	❑	❑
I may refer patients diagnosed with metabolic disorders added to the NBS panel to specialists in those disorders.	❑	❑	❑	❑	❑	❑
I am concerned about being reimbursed for all of the care that I would provide to patients diagnosed with a disorder on an expanded NBS panel	❑	❑	❑	❑	❑	❑
There are not currently enough practitioners educated to care for patients diagnosed with metabolic disorders on an expanded NBS panel	❑	❑	❑	❑	❑	❑
I am concerned that there would not be evidence-based treatment protocols to care for children diagnosed with disorders added to the NBS panel	❑	❑	❑	❑	❑	❑
Adding disorders to the NBS panel will make working with parents much more difficult	❑	❑	❑	❑	❑	❑
I am excited to think about being able to treat children with metabolic diseases that would otherwise have not been detected at birth	❑	❑	❑	❑	❑	❑
I am concerned that adding disorders to the NBS panel would result in a greater number of false-positives	❑	❑	❑	❑	❑	❑

Section 6: Best Practices

70. **This survey has asked you about processes of care and challenges to those processes for patients diagnosed with a metabolic disorder after a positive NBS and their families from the time of the screening through the first year of life. Thinking about your experiences and practice, are there any models of care or other things that have improved the care that patients and their families receive that you would like to share with us?**

_____

_____

_____

71. **In this survey, we asked about the educational needs of patients' families and primary care providers.**

How would you rate your need for additional education about metabolic disorders for the care of patients from birth to one year old?

- ❑ I do not have any additional education needs.
- ❑ I could use periodic updates about metabolic disorders and care practice.
- ❑ I would appreciate frequent updates about metabolic disorders and care practices.

72. **What mode of education do you find most helpful?**

- ❑ I prefer to research information myself on the web
- ❑ I prefer to have information mailed or emailed to me
- ❑ I prefer to receive information at conferences or professional meetings

Section 7: Wrap-Up

73. **Thinking about the topics asked about here and any other concerns you have regarding the care of children with metabolic disorders and their families during the first year of life, what is the greatest challenge you face in practice?**

_____

_____

_____

74. **What is the zip code of your primary practice location (for regional analyses only)?** _____

Debriefing Sheet

Thank you for completing the survey! This page will further explain the purpose of the survey research you have just participated in. After you are finished viewing this page and have submitted your answers by clicking on the button at the bottom of the page, it is recommended you exit or quit your Web browser to eliminate the possibility (which varies depending on your computer and browser) that your responses could be viewed by hitting the "back" button.

We would like to remind you that we will keep the information you provide confidential. Further, any potentially identifying information (e.g., zip code) will be used only for regional analysis.

Because you have invested time in this study, you may have an interest in what we hope to find from your results. The purpose of this study is to examine the process of care for patients diagnosed with metabolic conditions that detected on newborn screen (NBS) from diagnosis to the first birthday. This study also examines the challenges and complexities of providing care to these patients and their families.
If you have questions about this survey or would like a copy of the results when they are available, please email me or call me at the number below.

Your participation, and that of your colleagues, is important for influencing ongoing policy discussions regarding the importance of genetic medicine.

Thank you again for your interest and participation.

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Appendix B. Methodology Report

This appendix contains a brief summary of the dates and modes of survey distribution through participating organizations and networks as well as information on the final sample size.

Survey Distribution.

The survey was distributed to potential respondents in two phases. In the first stage, participants were recruited via electronic contact through the Genetic Service and Newborn Screening Regional Collaborative Groups (RCs). The RCs were able to contact members of their listserv and those who received official RC newsletters. Some RCs decided to work with local provider organizations and state NBS representatives in order to distribute the survey to the appropriate providers. Due to privacy concerns, RCs did not want to share their provider lists with provider staff. Because of lackluster response rates, a second stage of survey was employed. During this stage, an effort was made to recruit providers directly through provider organizations, like the Society for the Inherited Metabolic Disorders (SIMD) and the National Society of Genetic Counselors (NSGC). The method and dates of distribution are indicated below.

Table Appendix B. Date and Method of Survey Distribution by Organization

Organization	Date	Method
New England Genetics Collaborative (NEGC)	10/18/11 (via New England Consortium of Metabolic Programs)	Email to Consortium Planning Group members
	10/28/2011	eNewsletter
New York-Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC)	10/6/2011	Email
	1/20/2012	Email
Southeast NBS and Genetics Collaborative (SERC)	1/9/2012	email
Region 4 Genetics Collaborative	10/17/2011	Email
Heartland Regional Genetics and Newborn Screening Collaborative	12/7/2011	Email to providers in 8 states
	1/10/2012	Email to additional providers
Mountain States Genetics Regional Collaborative Center (MSGRCC)	9/30/2011	Newsletter
	10/19/2011	Email
	11/30/2011	Newsletter
Western States Genetic Services Collaborative (WSGSC)	10/7/2011	Email to select group of providers
	11/15/2011	Email to providers contacted 10/7/11
	11/21/2011	Email to regional dietitians and genetic counselors

National Coordinating Center for the Regional Genetic and Newborn Screening Service Collaboratives (NCC)	12/16/2011	Newsletter
Pediatric and Clinical Genetics special interest group listserv of the National Society of Genetic Counselors (NSGC)	1/20/2012	Listserv
NBS mailing list (Newborn Screening Inquiry/Discussion List) (NBS@lists.UTSCSA.edu)	1/23/2012	Listserv
Society for Inherited Metabolic Disorders (SIMD)	2/8/2012	Email
American College of Medical Genetics (ACMG) Annual Clinical Genetics Meetings	3/27-31/12	Conference: flyer in conference bags, survey on conference computers, booth
Genetic Metabolic Dietitians International (GMDI) Educational Conference	4/19-20, 2012	Conference: booth

Sample Size.

A total of 181 respondents viewed the survey. Of those 181, 35 reported no information on the survey, 3 reported only one piece of information, and 5 reported 2 pieces of information. Because they reported so little information, the decision was made to drop them from analyses. Additionally, 21 people who reported that they did not treat children diagnosed with metabolic conditions following a positive newborn screen were also removed from the sample. Two additional cases were removed from the sample because the respondents reported a zip code outside of the United States (Canada). The final sample size of usable reports for this report is 114.

Appendix C. Tables

Table 5. Challenges to Providing Care During the First Year of Life (n=84)[1]

	Very Challenging	Somewhat Challenging	Not Very Challenging	Not At All Challenging	Don't Know/ Not Available	Not Reported
Geographic Distance Between Providers and Patients/Families	42.86 (36)	51.19 (43)	4.76 (4)	-	-	1.19 (1)
Parental/familial compliance with treatment and management protocols	33.33 (28)	54.76 (46)	9.52 (8)	1.19 (1)	-	1.19 (1)
Language Barriers between Providers and Patients/Families	17.86 (15)	52.38 (44)	22.62 (19)	4.76 (4)	-	2.38 (2)
The Adequacy of Staffing at Your Clinic	17.86 (15)	35.71 (30)	33.33 (28)	8.33 (7)	1.19 (1)	3.57 (3)
Unique Protocols of Insurance Providers	35.71 (30)	41.67 (35)	13.10 (11)	1.19 (1)	4.76 (4)	3.57 (3)
Availability of Prescription Medications and/or Medical Foods	32.14 (27)	54.76 (46)	8.33 (7)	2.38 (2)	1.19 (1)	1.19 (1)
Ability to Transition Patients to Hospital Providers when they are Admitted	4.76 (4)	27.38 (23)	46.43 (39)	15.48 (13)	4.76 (4)	1.19 (1)
Having to Take on Other Roles When Providing Care to Patients/Families	13.10 (11)	52.38 (44)	26.19 (22)	7.14 (6)	1.19 (1)	-
Parent/Family Level of Knowledge and Awareness of their Child's Disorder	17.86 (15)	64.29 (54)	15.48 (13)	2.38 (2)	-	-
Your Own Educational Needs Concerning Metabolic Disorders Detected on NBS	5.95 (5)	28.57 (24)	38.10 (32)	23.81 (20)	2.38 (2)	1.19 (1)
Having Enough Time to Provide Patients/Families with All of the Resources They Need	40.48 (34)	44.05 (37)	11.90 (10)	2.38 (2)	1.19 (1)	-
Lack of Reimbursement for All of the Care that Patients/Families Need	61.90 (52)	27.38 (23)	4.76 (4)	1.19 (1)	3.57 (3)	1.19 (1)
Communication between the State NBS or Follow-Up Programs and Providers	3.57 (3)	15.48 (13)	35.71 (30)	41.67 (35)	2.38 (2)	1.19 (1)
Primary Care Providers' Knowledge of Metabolic Disorders	25.00 (21)	47.62 (40)	22.62 (19)	1.19 (1)	2.38 (2)	1.19 (1)

[1]Row percentages are shown.

Table 7a. Metabolic Perceptions of Primary Care Providers Participation in Patient Care (n=84)[1]							
	Strongly Agree	Agree Somewhat	Neither Agree nor Disagree	Disagree Somewhat	Strongly Disagree	Don't Know/ Not Applicable	Not Reported
General Perceptions							
Primary care providers (PCPs) should participate on metabolic patient care teams	44.05 (37)	28.57 (24)	16.67 (14)	8.33 (7)	-	1.19 (1)	1.19 (1)
PCPs often lack knowledge of the metabolic disorders detected on uniform screening panel	59.52 (50)	34.52 (29)	3.57 (3)	-	-	1.19 (1)	1.19 (1)
PCPs should act as the medical home for metabolic patients in the first year of life	39.29 (33)	25.00 (21)	15.48 (13)	14.29 (12)	1.19 (1)	2.38 (2)	2.38 (2)
PCPs should act as the liaison to the patient's insurance company	11.90 (10)	20.24 (17)	30.95 (26)	25.00 (21)	5.95 (5)	4.76 (4)	1.19 (1)
Communication with PCPs							
Communication occurs in both directions	21.43 (18)	38.10 (32)	9.52 (8)	20.24 (17)	4.76 (4)	-	5.95 (5)
Communication provides easy to utilize, factual information about the child's conditions, treatment, prognosis, medications, and co-morbid conditions	33.33 (28)	46.43 (39)	9.52 (8)	5.95 (5)	1.19 (1)	1.19 (1)	2.38 (2)
Communication with primary care providers is a priority	47.62 (40)	34.52 (29)	7.14 (6)	3.57 (3)	-	-	7.14 (6)

[1]Row percentages are shown.

Table 7b. Effectiveness of Methods to Share Patient Management with Primary Care Providers (n=84)[1]						
	Very Effective	Somewhat Effective	Not Very Effective	Not Effective At All	Don't Know/ Not Applicable	Not Reported
Periodic conference calls, including specialty and primary care providers, parents, others	16.67(14)	42.86 (36)	22.62 (19)	2.38 (2)	13.10 (11)	2.38 (2)
Active plan of care or action plan identifying current care needs, who will provide action, and when the action should be completed	39.29 (33)	47.62 (40)	3.57 (3)	-	8.33 (7)	1.19 (1)
Written emergency care plan	77.38 (65)	20.24 (17)	-	-	1.19 (1)	1.19 (1)
Consultation letter from the specialist describing findings, actions taken, and plans	63.10 (53)	28.57 (24)	5.95 (5)	-	1.19 (1)	1.19 (1)
Condition-specific “fact sheet” with key information about the condition	45.24 (38)	42.86 (36)	8.33 (7)	-	2.38 (2)	1.19 (1)
Shared or interoperable electronic health records	41.67 (35)	39.29 (33)	3.57 (3)	-	13.10 (11)	2.38 (2)
Regular email communication with primary care providers	23.81 (20)	47.62 (40)	14.29 (12)	1.19 (1)	11.90 (10)	1.19 (1)
Other	2.38 (2)	2.38 (2)	-	-	4.76 (4)	90.48 (76)

[1]Row percentages are shown.

Table 8. Attitudes Toward Expanding the NBS Panel (n=83)[1]							
	Strongly Agree	Somewhat Agree	Neither Agree nor Disagree	Somewhat Disagree	Strongly Disagree	Don't Know/ Not Applicable	Not Reported
Increasing the number of metabolic disorders on the NBS panel in my state would increase my caseload	60.24 (50)	32.53 (27)	6.02 (5)	-	-	1.20 (1)	-
Expanding the number of metabolic conditions on the NBS panel would increase the time I spend treating patients.	54.22 (45)	32.53 (27)	9.64 (8)	1.20 (1)	-	1.20 (1)	1.20 (1)
I may refer patients diagnosed with metabolic disorders added to the NBS panel to specialists in those disorders.	15.66 (13)	14.46 (12)	20.48 (17)	15.66 (13)	19.28 (16)	14.46 (12)	-
I am concerned about being reimbursed for all of the care that I would provide to patients diagnosed with a disorder on an expanded NBS panel	40.96 (34)	27.71 (23)	13.25 (11)	2.41 (2)	4.82 (4)	10.84 (9)	-
There are not currently enough practitioners educated to care for patients diagnosed with metabolic disorders on an expanded NBS panel	62.65 (52)	19.28 (16)	14.46 (12)	2.41 (2)	-	-	1.20 (1)
I am concerned that there would not be evidence-based treatment protocols to care for children diagnosed with disorders added to the NBS panel	40.96 (34)	30.12 (25)	14.46 (12)	12.05 (10)	1.20 (1)	-	1.20 (1)
Adding disorders to the NBS panel will make working with parents much more difficult	6.02 (5)	13.25 (11)	30.12 (25)	24.10 (20)	22.89 (19)	1.20 (1)	2.41 (2)
I am excited to think about being able to treat children with metabolic diseases that would otherwise have not been detected at birth	44.58 (37)	30.12 (25)	16.87 (14)	6.02 (5)	-	1.20 (1)	1.20 (1)
I am concerned that adding disorders to the NBS panel would result in a greater number of false-positives	32.53 (27)	31.33 (26)	18.07 (15)	8.43 (7)	6.02 (5)	2.41 (2)	1.20 (1)

[1]Row percentages are shown.

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