Newborn Screening Education: A Survey of Ontario Mothers

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Thesis submitted to the Faculty of Graduate and Postdoctoral Studies in
Partial fulfillment of the requirements for the MSc degree in Epidemiology

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ABSTRACT

Purpose and methods: Effective parental education about newborn screening (NBS) may help to maximize the benefits and minimize the harms of screening. We investigated experiences, knowledge and opinions regarding NBS education among Ontario mothers. Mothers whose infants recently received NBS were invited to complete a mailed survey (n=1712).

Results: Of the 750 participants, 93% recalled their infant receiving NBS, while 69% recalled receiving information about NBS. Of this group, fewer than 50% reported receiving information prenatally, yet a majority of mothers (64%) viewed this as the most important time for education. Those who received information prenatally reported higher satisfaction (OR 2.4). The 40% of mothers who recalled being informed about the meaning of results had higher knowledge about NBS (OR 2.7) and reported higher satisfaction (OR 4.2).

Conclusions: Parental education about NBS could place greater emphasis on the prenatal period and on fostering understanding about the meaning of results.

ACKNOWLEDGEMENTS

I would like to express my sincere appreciation to my supervisor Dr. Beth Potter for her continued support and guidance throughout the duration of this thesis. I would also like to thank Dr. Brenda Wilson and the other members of the thesis committee, including Dr. Pranesh Chakraborty, Christina Honeywell, and Tim Ramsay for their time and for providing invaluable guidance. I would also like to acknowledge Jennifer Milburn, Nicole Gaffney and Kim Gall for their patience and hard work. I would also like to thank Silvia Visentin for her assistance with survey logistics. I would like to acknowledge and thank the participants in this study. Finally, I would like to thank my family and friends for their continued support and encouragement.

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LIST OF ABBREVIATIONS

AAP- American Academy of Pediatrics

ACMG- American College of Medical Genetics

ACOG- American College of Obstetricians and Gynecologists

BIO- Biotindase Deficiency

CAH- Congenital adrenal hyperplasia

CAN- Canada

CH- Congenital hypothyroidism

CI- Confidence interval

CMES-Canadian Maternity Experiences Survey

GALT-Galactosemia

HCP-Healthcare professional

HRSA-Health Resources and Services Adminitration

INFO- Information

MCHB- Maternal and Child Health Bureau

NBS- Newborn blood spot screening

NNGRC- National Newborn Screening Genetics Resource Center

NSO- Newborn Screening Ontario

OR- Odds ratio

PKU- Phenylketonuria

US- United States

Chapter 1: Introduction

1.1 Overview of newborn screening

Newborn 'blood spot' screening (NBS) is a population-based public health initiative aimed at the early detection of infants at-risk for certain rare but serious disorders (primarily inherited).[1-5] NBS is recognized as an important public health tool for reducing morbidity and mortality among infants with disorders including inborn errors of metabolism, endocrine disorders, hemoglobinopathies and cystic fibrosis.[5-8] Population screening of newborns first began in the 1960s with the introduction of the Guthrie test for phenylketonuria (PKU).[2,5,6,8-10] NBS for PKU was first initiated by Guthrie's development of a bacterial inhibition assay which he used to detect elevated blood levels of phenylanine (a biomarker for PKU).[2,3,8,9,11,12] This discovery had major implications for population screening, demonstrating the capability for mass screening of samples which could be processed rather inexpensively.[2,8,9]

The NBS process requires that a sample of blood be taken from the infant during the first few days of life (usually no earlier than 24 hours after birth). Specimens of blood are typically collected using a heel prick and are absorbed onto filter paper and sent to a centralized laboratory for testing to identify markers indicative of a high risk for one or more specific disorders.[2,8,11,13] Testing strategies differ depending on the disorder, but positive (abnormal) results suggest a need for further testing to confirm or exclude a diagnosis. Infants who screen positive are referred to a treatment centre. Treatment centres notify the parents and/or guardians, sometimes through their family physician if available, and retrieve the infant for additional testing and treatment as necessary. Parents are not typically informed of negative (normal results) but can request that they be sent to their family doctor.

Depending on the results of confirmatory testing and/or symptom development, there are five possible types of result that may eventually arise from NBS: true positives, true negatives, false positives, false negatives and carriers. Most infants will have true negative results: the NBS test for each condition will be negative and the infant will never develop symptoms of the screened diseases. Very occasionally NBS leads to false negative results, whereby the NBS test is negative but the individual is diagnosed with the disease, often after developing symptoms. False negative results are rare (far less common than false positive results); in part due to testing methods that deliberately have extremely high sensitivity at the expense of specificity.[8] In the case of true positive results, an infant has a positive screening test and is shown through confirmatory testing to be affected with the disorder. A false positive result is one where an individual is identified as being at high risk based on the screening test but the disease is later ruled out. Finally, NBS for genetic conditions with autosomal recessive inheritance patterns may lead to the identification of carriers. A carrier is an individual who has only one copy of a disease-causing mutation and is therefore not affected with the screened condition (although s/he is at risk of passing the mutation to his/her own offspring and the carrier result means that one of the infant's parents is an obligate carrier).[14] Carrier identification is not a goal of NBS, but some babies will be identified as carriers by the screening and diagnosis process described above.

1.2 History

1.21 Newborn screening in North America

Screening for PKU in the US was first offered in 1962 by the Erie Department of Health in Guthrie's home state (New York).[9] During this time the Maternal & Child Health

Division (MCHD) of the US Children's Bureau also implemented a national pilot study of the PKU test to examine its feasibility for mass screening.[2,6,9] In 1963 Massachusetts became the first state to mandate testing of all newborns for PKU.[2,9] Guthrie continued to be a strong voice advocating NBS for PKU and by 1985 most state programs began pilot screening for PKU. [9]

NBS expansion was first initiated by the invention of the Phillips Punch Index Machine. This machine allowed for the extraction of multiple samples from a single blood spot, thereby substantially reducing the amount of effort required for multiple sample preparation.[9] Efforts to expand screening began with the development of a dried blood spot (DBS) screening test first for congenital hypothyroidism (CH) and later for various other conditions such as congenital adrenal hyperplasia (CAH).[2,9] Advances in computer technology led to the development and use of microcomputers in NBS for data management and follow-up. This facilitated the growth of programs that were otherwise restricted due to limited resources for data handling.[9]

NBS expanded further with the development of newer technologies such as tandem mass spectrometry (MS/MS). The introduction of MS/MS in the 1990s further improved screening efficiency through the ability to detect numerous conditions from a single blood spot, therefore making it possible for programs to screen newborns for up to 50 conditions using one blood spot.[5,7,15-17] The combination of automated punching, dried blood spot procedures, computer technology and MS/MS has resulted in the considerable expansion of many state programs to include numerous rare conditions such as: amino acid disorders, fatty acid disorders, organic acid disorders, certain hemoglobinopathies, endocrine disorders and cystic fibrosis (CF).

NBS for metabolic and heritable disorders in Canada developed in parallel with the US, with the first documented NBS program established in 1963 in Prince Edward Island.[9,15] By 1970 all other provinces except Newfoundland and Labrador had established NBS programs. Screening in most provinces focused primarily on PKU and CH from 1970s through the 1990s with the exception of Manitoba and Quebec which expanded their screening programs.[9,15] In addition to PKU, Manitoba initiated screening for other amino acid disorders, galactosaemia, fructose intolerance and began targeted screening for Duchenne muscular dystrophy (which has since been discontinued); while Quebec began screening for galactosaemia and tyrosinaemia in blood spots and screening for amino acid and organic acid disorders in urine.

Currently, all provincial NBS programs include PKU and CH at minimum. Among the provinces, the number of conditions included in routine screening currently ranges from six (Newfoundland and Labrador) to 47 (Saskatchewan).[18] Newborn screening in Ontario began in 1965. Efforts to expand screening began in 1978 with the addition of CH to the screening panel.[19] Further changes to the program did not occur until the announcement in November 2005 that the provincial government planned to expand the Ontario NBS program and relocate its services to Ottawa from Toronto.[19] Shortly following this announcement the Ontario NBS program was established at the Children's Hospital of Eastern Ontario and equipped with MS/MS technology. In 2006, the program began screening for 18 additional disorders, including certain fatty acid oxidation disorders, amino acid disorders, organic acid disorders and hemoglobinopathies.[19] Between 2007 and 2009, the program expanded further and began screening for biotinidase deficiency (BIO), galactosaemia (GALT), congenital hyperplasia (CAH) and cystic fibrosis (CF). Thus, NBS

in Ontario has expanded to include a core panel of 28 conditions. These 28 are the same conditions that were recommended by the American College of Medical Genetics (ACMG)[7,13] (See appendix 1, Table 24 for a list of disorders currently included in Ontario's NBS panel).

1.22 Current practices & controversies in newborn screening

Expanded NBS has been debated, particularly in terms of which conditions meet the criteria for screening.[1,7,20] Generally, screening practices are based on Wilson and Jungner's principles for appraising the validity of a screening program (Table 1). As a result, most jurisdictions recommend screening for diseases for which there are effective treatments that prevent and/or modify the progression of disease, and for which funding is available for follow-up and management. [2,6,9] However, evidence supporting the clinical benefits of NBS for some conditions included in expanded panels is sparse, likely as a result of the rapid progression of available technologies (i.e., evidence has lagged behind) and because the conditions are rare (making NBS programs challenging to evaluate with scientific rigor).[7,21-24]

Table 1: Principles of screening for disease[25]

1. The condition sought should be an important health problem
2. There should be an accepted treatment for patients with recognized disease
3. Facilities for diagnosis and treatment should be available
4. There should be a recognizable latent or early pre-symptomatic stage
5. There should be a suitable test or examination
6. The test should be acceptable to the population
7. The natural history of the condition, including development from latent to declared disease should be adequately understood.
8. There should be an agreed policy on whom to treat as patients
9. The cost of case findings (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole.
10. Case-finding should be a continuing process and not a 'once and for all' project

Currently, NBS protocols and practices vary considerably across jurisdictions and international NBS policies are lacking.[7,9] Because North American NBS programs are governed at the state or provincial level, protocols and practices in individual states/provinces sometimes reflect local politics, economics and culture.[3,7,9] In the US, the Health Resources and Services Administration (HRSA) funded the American Academy of Pediatrics (AAP) to convene a NBS Task force in 1999 to review the issues and challenges facing NBS programs. The task force published a report which described a national agenda for NBS systems;[1,5,9,14] and recommended greater uniformity among state programs.[6,7]

In turn, the recommendations of the AAP report led the Maternal and Child Health Bureau (MCHB) to commission the American College of Medical Genetics (ACMG) to develop national guidelines for state NBS programs.[7,9] The ACMG's recommendations, published in a 2006 report, have been influential internationally as well as in the US.[77] These include a set of 29 core conditions and 25 secondary target conditions recommended for inclusion in all state NBS programs.[3,7] The ACMG also recognized a need for a US national NBS identity which would oversee the various components of a NBS system, including data collection, program evaluation and education.[7] The recommendations of the ACMG and the process used in reaching them have been subject to debate and criticism. Critics of the ACMG group's recommendations question the scientific approach used in developing them. They argue that the process was based mainly on expert opinion rather than on empirical evidence and that there is a need to re-evaluate the recommendations.[26-29] Others, however, have endorsed the ACMG recommendations and have argued that the group used the limited published information available in developing their report.[30]

Since the ACMG report, a new US committee has been established and has developed a US national evidence and policy development process for NBS.[31,32] According to this committee, key principles influencing the process of selecting conditions to include in the uniform screening panel include a reliance on evidence, transparency, and consistency.[31,32]

To date there are no national guidelines or policies for NBS in Canada, although the ACMG recommendations appear to have been influential in some provinces, including Ontario. It has been argued that Canadian guidelines may help to improve provincial NBS practices and promote equity in access to services.[33]

Chapter 2: Literature review and study objectives

2.1 Importance and proposed benefits of NBS education

NBS is now recognized as a system of care (rather than an isolated test) that includes education, the screening test itself, short-term follow-up, diagnostic confirmation, management, and program evaluation.[4,7,11] It is considered important that each component of the system be synchronized so as to ensure that service gaps are not created.[7,34] This recognition of NBS programs as integrated health care systems has brought with it the need to address issues concerning the provision of various ancillary services, including pre-screening education.[7]

Effective pre-screening education for parents or prospective parents of infants in the screened population may help to ensure that they are aware of the possible risks of screening prior to testing, thus reducing the possibility of negative psychosocial outcomes, such as psychosocial harm from receiving either a true or false positive result.[3,23,35,36] Another proposed benefit of educating parents about the NBS process is the potential for improving follow-up rates for children identified as needing additional testing due to poor quality samples or positive screening results by providing parents the opportunity to understand the benefits, purpose, and process of screening.[37]

A majority of US and Canadian NBS programs operate on a mandatory (US) or an "implied consent" (Canada) basis.[1,38] In the past, implied consent for screening for PKU and CH was not typically contested because screening for these conditions was recognized as highly effective for preventing severe disability.[21,22,38] However, with expanded screening this practice has been challenged, since the balance of benefits and harms of early identification and/or treatment for some conditions are less clearly defined (as described

earlier, clinical effectiveness evidence is scarce for some conditions included in expanded NBS).[21,22,38] Thus, some have acknowledged a "tension" between promoting informed choice and supporting the public health goal of screening.[12,38] It has been argued that the provision of accurate, clear information and the use of effective communication strategies between parent and provider may help to relieve this tension,[12,38] although the complexity and number of diseases included in an expanded screening panel may create challenges in providing clear information.

Finally, in the context of debate and controversy about expanded NBS, transparency in decision making via high quality communication with parents and the public may be an important way of ensuring procedural fairness in policy decision-making.[39-42] For example, transparency has been a focus of discussion with respect to policies about the storage and secondary use of residual dried NBS blood samples.[39,40]

2.2 Guidelines for NBS education

Differences have been observed across NBS programs in terms of pre-screening education, which may be in part due to a scarcity of relevant guidelines.[5,12] As mentioned, there are no Canadian national guidelines covering aspects of NBS policy, including education. Here the US and UK guidelines for parental NBS education are briefly reviewed.

In the US, the 1999 report by the US NBS Task Force made several recommendations for improvements within the area of education including timing and content of education.[6] The ACMG report also discussed educational practices and further US guidelines have been focused specifically on NBS education.[7,22,43] In the UK, NBS policy is currently coordinated at the national level, although there are some differences in screening panels across the four

UK countries.[44] The UK NBS program has developed specific guidelines/recommendations for NBS education including when education should take place and what information should be included in discussions with parents.

With respect to timing of parental education about NBS, the prenatal period has been endorsed by the AAP, ACMG and American College of Obstetricians and Gynecologists (ACOG) as the optimal time for parents to receive information about NBS; [6,7,22] the UK Newborn Screening Programme Centre also recommends providing a pre-screening leaflet prenatally.[44] The ACMG report further recommends that education should occur at multiple times throughout the screening system, and should involve discussion between parents and healthcare providers.[7]

In addition to addressing the timing of education, the AAP task force also made recommendations for the type of information that should be included in parental discussion about NBS, which include the following: 1) benefits of screening, 2) potential risks of the test, 3) how parents are informed of results, 4) possibility of false-positive results, 5) importance of responding to a positive result, 6) how to respond to a positive test, and 7) the screening program's policy for sample storage and use of stored samples.[6,45]

More recent US-based guidelines for the content of NBS communication have been developed through an evidence-informed process involving a review of existing brochures and literature and focus groups with parents and professionals.[43] The resulting model brochure outlines a set of seven key messages that parents consider to be important to include in discussions about NBS (Table 2).[43]

Table 2: Seven things parents want to know about NBS

Seven Things Parents want to know about NBS[43]
1- All newborn babies are required by the State to get tested for some rare disorders before they leave the hospital
2- Babies with these disorders may look healthy at birth
3- Serious problems can be prevented if we find out about the disorders right away
4- To do the test, a nurse will take a few drops of blood from your baby's heel
5- Your baby's heath professional and hospital will get a copy of the test results. Ask about the results when you see your baby's health professional
6- Some babies will need to be retested. If your baby needs to be retested you will be notified. It is very important to get retested quickly.
7- Talk to your baby's health professional if you have questions

The UK recommendations for the content of NBS education are presented in Table 3. Notably, these guidelines recommend informing parents about the screened conditions and about the possibility of carrier status results. Fewer conditions are included in UK NBS programs relative to most North American programs, which may in part account for the recommendation to specifically discuss the screened diseases.

Table 3: UK screening programme's guidelines for communication

Information to be included in discussion with parents about NBS[44]
• Conditions and how screening can help infants with these conditions
• How the blood sample is taken, and that sometimes a second sample is needed
• When parents should receive results
• Screening for Sickle cell and Cystic Fibrosis can identify babies who are carriers
• Screening is not 100% accurate

While there are no specific guidelines for who should be responsible for providing pre-screening education in the US and Canada, the endorsement of the prenatal period implies that prenatal healthcare professionals (e.g., obstetricians and family physicians) should provide this information.[6,7,22] According to the ACOG, pre-screening education could

be provided at the first obstetric visit (the NBS pamphlet could be included with other information distributed at this time);[22] in the UK recommendations a leaflet is to be provided during the third trimester of pregnancy by midwives (who are the primary prenatal caregivers in the UK).[44]

2.3 Empirical evidence

There is an awareness of the need for further research into communication practices with parents about NBS.[12] Specifically, limited research has examined parents' and professionals' views and experiences with respect to pre-screening education.[12] We were able to identify eight recent studies that empirically examined parental attitudes and/or experiences with NBS education.

Campbell et al. explored parental attitudes and beliefs concerning genetic screening. In four out of 13 focus groups (31%) no parent was able to recall any details of NBS. Further, parents in seven out of 13 focus groups (53%) spontaneously suggested that education about universal NBS should take place prior to delivery, and that it [education] should be incorporated into routine prenatal care.[46] Similarly, a study conducted by Davis et al. which involved 22 focus groups and three individual interviews also found that parents had limited knowledge /awareness about NBS practices. All parents agreed that the worst time to receive information was during hospitalization, as this proves to be the most distracting time for a parent, and that the optimal time for education would be during the third trimester of pregnancy.[43] A more recent study by Tluczek and colleagues involving interviews with 193 parents reported similar findings.[47] Most parents in Tluczek's study reported having very little knowledge about NBS, and six percent indicated that they did not recall receiving any

information at all.[47] With respect to timing of education, most parents reported receiving information either during hospitalization (26%) or during specimen collection (12%).[47] Additional studies also illustrate that parents are generally unaware of NBS and that most often they are being informed about NBS close to when the heel prick is performed, despite expressing support for provision of information prenatally.[48-52] For example Suriadi and colleagues interviewed Australian mothers whose infants recently participated in NBS (n=200) about their knowledge about NBS and found that very few (27%) mothers were aware of NBS.[48] Davey et al. surveyed Australian mothers (n=600) on their knowledge of NBS and their opinions on the storage and use of blood samples.[49] In contrast to the findings in other studies, the majority of mothers in this study (93%) had heard of NBS. Most mothers, however, recalled receiving information after the birth of their child, and felt that the timing of education was a significant factor impeding their ability to process the information.[49] Detmar et al. conducted focus groups with parents and prospective parents to investigate their preferences and attitudes towards NBS education, as well as their views on consent and NBS expansion. Most participants in this study could not recall receiving any information about NBS. Very few mothers agreed with the timing of education and expressed their preference for receiving information during pregnancy.[50] Similarly, mothers of newborns who recently participated in NBS in Wales were recruited to take part in semi-structured interviews (n=18), with the purpose of exploring mothers' experiences with NBS.[51] According to Parsons and colleagues, the majority of mothers (83%) reported receiving information about NBS postnatally. Furthermore, Lang et al. interviewed mothers living in the Chicago area (n=388) with the purpose of investigating their understanding of and attitudes towards NBS.[52] The investigators found that few mothers (35%) recalled their

prenatal provider mentioning NBS, whereas 56% indicated being told about NBS at the hospital (in the nursery).[52] The results of studies of providers or NBS professionals have also shown that parents are more likely to receive education postnatally.[21,37,53-55]

With respect to the manner in which parents are educated about NBS, parents most often report receiving information either through conversation with a healthcare professional at the hospital or from an information brochure/pamphlet.[43,47,49-52]

Two studies have explored parental views about the information they feel is most important to include in discussions about NBS with parents. According to Davis et al. parents expressed a desire for basic information such as: 1) that screening would occur, 2) screening is beneficial, 3) the infant may require retesting, and 4) how results would be communicated.[43] Similarly, parents in the study by Tluczek et al. were interested in receiving information about; 1) the purpose of screening, 2) the importance of having your infant tested, 3) conditions included in screening, and 4) when and how results would be communicated.[47]

We identified four studies that have examined NBS educational materials for parents. One such study was conducted by Fant and colleagues and looked at the completeness and complexity of information available for parents from NBS programs in the US[45]. The authors found that, while 46 out of 47 (98%) of state programs reported providing standardized materials for parents, none of the program materials they obtained contained more than five of the seven elements recommended by the AAP.[45] Similarly, a study conducted by Arnold et al. found that in a majority of brochures the essential information for parents was buried in lengthy text including less relevant information.[56] They suggested that 81% of program materials needed improvements.[56] Similarly, in an international study

which looked at written information available to the public (through various sources) in Canada, the US, UK and Australia, Hargreaves and colleagues found that the majority of educational materials included information on the purpose, benefits and process of screening, whereas few addressed the potential harms (e.g., false positive results), consent process or when/how results would be disseminated.[57] In a more recent review of NBS education materials accessible from North American NBS websites, which was initiated in preparation for this thesis project, findings concur with previous studies that there is considerable diversity across programs in terms of the content of education materials, with little attention to the potential harms of screening in particular.[58]

2.4 Summary

In summary, there is general agreement that pre-screening parental education is an important part of NBS programs. Pre-screening education may be based on established guidelines (informed by research and/or based on professional consensus) or on empirical research identifying the elements of screening that are most important in terms of parental priorities, and in terms of maximizing the benefits of screening (for example, encouraging prompt follow-up for positive results), and minimizing its harms. The review provided above suggests that guidelines for pre-screening education do exist but are often not followed and may not be based on strong empirical evidence, and that to date primary research to determine parental priorities for education includes only a few, mostly small and qualitative, studies. In addition, to date, most of the published evidence concerning parental opinions/experiences of NBS has resulted from research conducted in the US and the UK and there are no national standards or guidelines for system performance in Canada. The

recent expansion of NBS programs has seen the inclusion of some disorders for which the effectiveness of early identification and/or treatment are not well defined. NBS expansion has also created challenges for parental education: a lack of knowledge about the conditions being tested, the number of conditions covered, and limited time may impede providers' ability to discuss NBS with parents. The available evidence suggests that generally parents are not well-informed about NBS.

2.5 Newborn screening education in Ontario and rationale for this study

Currently Newborn Screening Ontario (NSO) uses various methods to provide information to parents and prospective parents about NBS. For example NSO has developed a 'tear-off sheet' that is attached to the filter paper card used to obtain the NBS blood sample. The tear-off sheet can be given to parents at the time of the heel prick. This sheet describes the purpose of the screening test, how results are communicated, the meaning of positive and negative results, the right to refusal and that repeat testing may be required (e.g., if the sample was taken before 24 hours). Parents and prospective parents (as well as providers) may also receive information through the NSO website. Information for parents on the website is quite similar to that which is provided on the tear-off sheet but provides slightly more detail (for example how the sample is collected, and information about the conditions screened). The NSO website also provides parents with the option of downloading pamphlets from the Ontario Ministry of Health and Long-term Care, available in various languages.

In light of the recent expansion of the NBS program in Ontario, NSO program coordinators recognized a need for ongoing evaluation and needs assessment in the area of

parental education. This thesis project is the first study in Canada to empirically examine mothers' experiences with NBS and their views on how best to educate parents about NBS. This is the first study, to our knowledge, in Canada and the US to explicitly evaluate parental knowledge about NBS and explore potential predictors of having high knowledge and high satisfaction with NBS education. The insights gained from this study will make an important contribution to the research literature and will be useful to NSO, by informing the development and/or modification of education initiatives.

2.6 Goals and objectives

The purpose of this study was to investigate experiences, knowledge and opinions regarding newborn screening education among a representative sample of Ontario mothers, and to quantify associations with individual factors that may influence these variables. We aimed to generate knowledge useful to NSO for the development and/or modification of education initiatives as part of the program's ongoing evaluation and quality assurance process. Moreover, we hoped to make an important contribution to the small body of existing research literature on this topic. We chose to focus on mothers in part because they are most likely to have a first-hand encounter with NBS education. More specifically, it is likely that mothers (more than fathers) would be targeted in education strategies about pregnancy, childbirth and newborn care.

The specific objectives of this study were to:

1. Identify where Ontario mothers currently receive general educational information about pregnancy, childbirth and newborn care in order to inform potential targeting of NBS education strategies.

2. Describe Ontario mothers' experiences with education about NBS specifically (i.e.: to understand when, from whom, how and what information they received prior to or during screening).
3. Describe mothers' opinions about how best to educate Ontario parents about NBS (i.e.: their views on when, from whom, how and what information would be most helpful).
4. Develop a knowledge score and use this to ascertain mothers' knowledge about NBS
5. Explore important differences in mothers' experiences with, and knowledge and opinions about NBS education with respect to key demographic characteristics (e.g., age, education, language most often spoken at home and parity).
6. Identify aspects of mothers' experiences that are associated with knowledge and satisfaction regarding NBS education.

With the recent expansion of NBS programs, pre-screening education for parents is a relatively new area of research. Given that there were few previous studies on this topic, and none to our knowledge that empirically examined mothers' knowledge, satisfaction and opinions about NBS education, our approach was exploratory. Thus there were no pre-determined hypotheses to be forwarded or tested. Rather, the findings of this study have led to the development of potential hypotheses which could be explored in future studies (refer to section on future directions).

Chapter 3: Methodology

3.1 Study design

This descriptive study used a population-based survey to gather cross-sectional data from a random sample of mothers whose infants had recently participated in newborn screening in Ontario. We chose to use a self-administered questionnaire which was mailed to selected mothers.

3.2 Sample size

When estimating the sample size for a study with the goal of making inferences about a population proportion one must provide a preliminary estimate of the proportion to use in the sample size formula. One option is to use an estimate from a previous (similar) study. A second option involves using an estimate based on pilot data. In our study these options were not available: our objectives were multi-faceted (no single estimate was of primary interest) and similar studies do not exist in the literature. When these options are unavailable for estimating sample size a third acceptable option involves using p=0.50, which would produce the most conservative sample size estimate.[59] The sample size for this study was determined using the following formula for sample size needed to estimate a population proportion (see appendix 2 for calculations):[59]

$$n = \frac{p(1-p)[Z_1 - (\alpha/2)]^2}{E}$$

Where $Z_1-(\alpha/2)$ reflects the desired level of confidence (1.96)
p= the population parameter (0.50)
E= margin of error (0.50)

A sample size of 385 mothers was calculated using this approach. A study by Etchegary et al which surveyed Ottawa mothers about their knowledge and attitudes toward genetic testing

reported a low (41%) response rate.[60] We assumed a 50% response rate and so the sample size of 385 was doubled for a sample of 770. It was estimated a priori that it may be useful to stratify some estimates by parity (mothers who had only one child vs. those with multiple children). The Canadian Maternity Experiences Survey (CMES) reported that 45% the women in their sample had one child at the time of the study.[61] Based on this the sample size was further inflated to ensure that there were at least 770 sampled from each group. Therefore we estimated that the final sample size required was 1712 mothers (refer to appendix 2 for detailed sample size calculations).

3.3 Recruitment

The NSO database was used as a sampling frame for this study. This database includes the names and contact information of mothers for all screened infants along with their infants' screening results. Mothers were considered eligible for inclusion in the study if their youngest child participated in the NBS program, was six months or younger at the time of the study and received a negative screening result. Mothers were excluded if their infant:

a) Was older than 6 months at the time of the study
b) Received a positive screening result
c) Was adopted
d) Required repeat testing
e) Had an unsatisfactory result
f) Was deceased

Mothers were also excluded if there was missing contact information in the NSO database or if they had duplicate records in the database.

In the first step of sample selection, a NSO staff member (JM) conducted a query of the NBS database to identify mothers of all infants born between April 1, 2009 and May 31, 2009. Mothers who were deemed ineligible based on the above criteria were excluded from the study. A database in Microsoft Excel was created with a list all eligible participants (n=23,386). A random number generator was used with uniform distribution and parameters from 0-1 to create a new column with random numbers. The database was sorted by ascending order using the random number column with the first 1712 records selected for the study sample.

The NBS program contacted parents up to five times to invite their participation in the study following Dillman's evidence-informed "tailored design method".[62] Table 4 lists the documents included in each mail-out, and the date each package was mailed.

Table 4: Description of Mailings

Mail-out	Documents included in the package	Date
1	Pre-notification letter	July 23, 2009
2	Cover letter, questionnaire, participant information form, $2 Tim Hortons' gift card	August 3, 2009
3	Reminder post-card	August 10, 2009
4	Cover letter, questionnaire, participant information form	August 26, 2010
5	Cover letter, questionnaire, participant information form	September 16, 2010

All questionnaires were accompanied by a detailed cover letter and information sheet explaining the purpose of the study and the importance of responding[62] and including additional information in accordance with the Children's Hospital of Eastern Ontario research ethics board guidelines (see appendix 3 for examples of each cover letter, and participant information form). To increase response rates and reduce non-response bias we

included a $2 Tim Hortons' gift certificate as an incentive with the second mail-out.[62] With each contact, mothers were informed of how they were selected into the sample, and assured that researchers outside the NBS program were not given their information. Potential participants were told that they could choose to participate in the study by filling out the questionnaire and sending it directly to the NBS program using a business reply envelope or by completing the questionnaire online on a secure website. Mothers who did not wish to participate were prompted to: a) return the blank questionnaire in the postage-paid envelope provided, b) call the NBS program to ask to be removed from the study mailing list, or c) simply ignore the five contacts and they would not be contacted further about the study. The questionnaire included an ID number to allow the NBS program to track responses and remove people from the mailing list as appropriate. The questionnaire did not, however, include any names or contact information. Researchers outside the NBS program received de-identified questionnaires (with ID numbers). Mothers who wished to participate in the study could mail a completed questionnaire to the researchers. We also provided participants the option of completing the survey online. Each potential participant was provided with a unique username and password and the website address, allowing mothers to securely access the questionnaire. The online questionnaire was designed to closely resemble the paper form. Participants could return to the online questionnaire multiple times if needed.

3.4 Instrument design

Because we were unable to identify any previous survey research on the topic of NBS education the questionnaire for this study was developed by MA with guidance from members of the thesis advisory committee. A complete draft of the questionnaire was reviewed by all members of the advisory committee and by some additional genetic counselors and investigators involved with the

NBS program. A pilot test of the study questionnaire was also conducted with a smaller sample of new mothers (see below for more details).

The questionnaire consisted of five sections and was structured so that each survey question was linked to one of the six study objectives (refer to appendix 4 for a copy of the study questionnaire). For example, with respect to the first objective (general education information about pregnancy and newborn care) questions addressed the following:

- Sources of information about pregnancy/child care (e.g., prenatal care provider, prenatal classes, friends and family, books or other educational material, etc).

In order to meet the second objective (mothers' experiences with NBS education) questions were asked concerning:

- Awareness of newborn screening
- Timing (e.g., prenatally vs. post-natally)
- Sources of NBS education (e.g., prenatal care provider, family other health care professional, prenatal class, pamphlet distributed in hospital) and;
- Content of information received (e.g., the purpose of screening, screening process, how results would be communicated, the meaning of positive and negative results, policies on sample storage and use).

The third objective (describe mothers' opinions about how best to educate parents) was achieved by asking mothers the following:

- Who mothers expect to be the most knowledgeable about NBS
- When mothers feel is the best time for parents to receive information about NBS
- How mothers feel parents should receive information
- What messages mothers feel are important to include in education to parents.

Mothers were also asked a series of five questions (Questions 25-29 in section 4, see appendix 4) to probe their knowledge about NBS (as per objective 4). These questions were developed following discussion with NSO staff and were considered to be important information for parents to know. They included questions about the purpose and timing of NBS, how results would be communicated, the meaning of a positive result and the right to refusal (see appendix 4). Additionally, mothers were asked to provide demographic information (e.g., age, gender, language of origin), to enable investigation of differences across these characteristics (as per objective 5). To achieve the sixth objective mothers were also asked to indicate their level of satisfaction (on a 5-point scale) with the information they received about NBS.

3.5 Pilot study

We conducted a pilot test of the questionnaire by visiting prenatal classes at the Ottawa Hospital (the pilot study received ethics approval from the Ottawa Hospital Research Ethics Board, appendix 5). Expectant mothers were approached by investigators (MA, BP) during three prenatal classes at the Ottawa Hospital. All packages included an information sheet about the study and two questionnaires: the main study questionnaire and a second shorter questionnaire that was used to inquire about pilot participants' opinions about various aspects of the main questionnaire (see appendix 5 for copy of pilot questionnaire and participant information form). Participants were asked to indicate on a four point scale their level of agreement to statements about:

- Clarity/comprehensiveness of questions and instructions
- Overall attractiveness of questionnaire
- Question flow and sequencing

- Length of time to complete

According to the wishes of the prenatal class instructors, all potential pilot participants were provided with a postage paid envelope to be used to return completed questionnaires after completion at home. No follow-up contacts were incorporated into the pilot study.

3.6 Data cleaning

All responses from each mailed questionnaire were recorded separately by two independent clerks, creating two (duplicate) datasets. Data entry clerks were provided with a list of codes for all responses to each question in the questionnaire. Clerks were instructed to 'flag' responses that they found difficult to code (e.g., where the clerk(s) could not read a response). All flagged questionnaires were reviewed and a list was compiled outlining the number and types of problematic responses. Issues were resolved by MA, and changes were incorporated into the final dataset. Discrepancies in data entry were identified first by merging together the two datasets from both clerks and then by carrying out a series of comparisons on all variables for each participant across both data sets. Where differences in coding were deemed to be a result of keying errors responses were corrected. Discrepancies which reflected real differences in coding were reviewed by checking against original responses in the questionnaire. A tracking system was used to identify the number of 'key-punch errors' for each clerk. The key error rate (KER) was calculated separately for each clerk using the following formula:

$$\text{KER} = \frac{\text{Number of surveys containing at least one key-punch error}}{\text{Total number of surveys entered}}$$

The dataset containing the fewest number of 'key-punch errors' was selected as the final dataset to be used for further analysis and all identified errors in this dataset were corrected. Responses from the web survey were merged with this dataset to create a final dataset to be 'cleaned'. A frequency count on all levels of all variables was conducted to identify potential errors. Responses for each variable were reviewed (by cross checking with list of codes) and problematic responses were recorded. A protocol was developed that outlined a series of quality checks to be taken to ensure that the data were 'clean', for example, checking for out-of-range responses, missing data, and incorrect responses (see appendix 6).

3.7 Analysis

Data analysis was carried out using SAS version 9.1 (University of Ottawa). All analyses were determined a priori. The analysis strategy was divided into four parts as follows:

Part 1:

To meet study objectives one through three, descriptive analyses (calculating proportions and confidence intervals) were carried out for all variables in the first three sections of the study questionnaire.

Part 2:

To address objective four, we developed a knowledge score based on the five knowledge questions (Questions 25-29) in the fourth section of the study questionnaire. We conducted descriptive analyses separately for each knowledge question and we determined the proportion of mothers with high (≥ 4 correct answers) versus low (0-3 correct answers).

Part 3:

We conducted binary logistic regression to look at the associations between demographic variables (age, parity, education, and language most often spoken at home) and the following variables (as per objective 5):

Experiences with NBS:

(i) Whether mothers recall receiving information about NBS (Q10)

(ii) When mothers recall receiving information about NBS (Q11- prenatal vs. postnatal).

(iii) Sources of NBS information (Q12) - We looked at demographic variables (age, parity, language and education) against all 12 categories (yes vs. no for all sources).

(iv) What information mothers recall receiving (Q14) - We looked at demographic variables (age, parity and education) against five categories for Q14 (1- purpose/benefits of NBS, 2- how screening would be done, 3- whether/how they would be told about positive/negative results, 4- the meaning of positive/negative results and 5- policy information on sample storage/use).

Opinions about NBS:

(i) When is the best time to receive information about NBS (Q17 –we broke this down into a dichotomous variable, prenatal vs. postnatal)

(ii) Important ways to provide this information (Q21- all six categories)

Part 4:

To address objective six a series of bivariate and multivariate analyses were conducted to look at mothers' experiences and opinions about NBS education.

(i) Knowledge

With respect to knowledge, we conducted bivariate analyses to explore relationships with each knowledge question separately and with the knowledge score (dichotomized high vs. low). The following variables were explored as predictors of knowledge:

a) Whether mothers recall receiving information about NBS (Q10)
b) Whether mothers received information prenatally vs. post-natally (Q11)
c) What information mothers recall receiving (Q14 in 5 categories)
d) Whether mothers recall receiving information from a healthcare professional(s) (Q12- as a dichotomous variable, yes vs. no/don't know)
e) Whether mothers recall receiving information from a prenatal class (Q12- as a dichotomous variable, yes vs. no/don't know)
f) Whether mothers recall receiving information from a tear-off sheet given at the time of the heel prick (Q13- as a dichotomous variable, yes vs. no/don't know)
g) Demographic characteristics (age, parity, education, language – English vs. other)

Multiple logistic regression analyses were used to construct models. Backwards elimination was used to test the significance of variables included in the models. We used SLSTAY=0.05 to specify the significance level for removing variables, and so variables were removed if their associated significance level was greater than 0.05. In multivariate analyses, we focused on the knowledge score as a dichotomous variable. Two models were constructed because Q10 applied to the full sample whereas Q11 and Q14 were only asked of mothers who reported having received information about NBS:

Model 1: Predicted mothers' knowledge (as a dichotomous outcome variable high vs. moderate/low) with the following possible independent variables: age, education, parity, language, Q10 (whether mothers recall receiving information about NBS, 2 levels).

Model 2: Predicted mothers' knowledge as a dichotomous variable with the following possible independent variables: age, education, language, parity, Q11 (whether mothers recall receiving information prenatally vs. post-natally), Q12 (whether mothers recall receiving information from a health care professional and/or from a prenatal class, 2 levels), Q13 (whether mothers recall receiving information from a tear-off sheet given at the time of the heel prick, 2 levels), and Q14 (what information mothers recall receiving; in 5 categories).

(ii) Satisfaction

We conducted bivariate analyses, to explore relationships with satisfaction (Q15) by including all five levels of satisfaction and by coding it as a dichotomous variable ("very satisfied" or "somewhat satisfied" versus all other levels of satisfaction). The following variables were explored as predictors of satisfaction:

a) Whether mothers received information prenatally vs. post-natally (Q11)

b) What information mother recall receiving (Q14 in 5 categories)

c) Whether mothers recall receiving information from a health care professional (Q12, 2 levels)

d) Whether mothers recall receiving information from a prenatal class (Q12, 2 levels)

e) Whether mothers recall receiving information from the tear-off sheet (Q13, 2 levels)

f) What information mothers recall receiving (Q14 in 5 categories)

g) Demographic characteristics (age, parity, education, language – English vs. other).

When conducting multivariate analyses, we focused on satisfaction as a dichotomous variable:

Model 3: Predicted mothers' satisfaction (as a dichotomous variable- satisfied vs. neither satisfied nor dissatisfied/somewhat dissatisfied/not satisfied) with the following possible independent variables: age, education, language, parity, Q11 (similar as 'model 2'), Q12 (same as 'model 2'), Q13 (same as 'model 2) and Q14 (same as 'model 2').

(iii) Other relationships among aspects of experiences and opinions

a) We examined the bivariate relationship between variables representing whether mothers received information prenatally vs. post-natally (Q11) and their opinions about when is the best time to receive this information (Q17).

b) Finally, we examined the bivariate relationship between variables representing what information mothers recall receiving (Q14) and their opinions about what information parents should receive (Q23).We examined the type of information received (Q14) in five categories (1- purpose/benefits of NBS, 2- how screening would be done, 3- whether/how they would be told about positive/negative results, 4- the meaning of positive/negative results and 5- information about sample storage/use) against Q23 (what information mothers feel is important for parents to receive) in the same five categories.

3.8 Ethical considerations

In accordance with Ontario privacy legislation and research ethics guidelines, all contacts with potential participants were made by a legitimate authority from Newborn Screening Ontario (see letter of support from the program, appendix 7). Appropriate procedures were in place to protect the confidentiality of study participants. These included the use of codes rather than identifiers and storing the master list linking names and codes securely and separately. Moreover, to further ensure patient confidentiality, researchers not involved in the NBS program did not have access to the contact information in the NBS database.

Since we did not have access to health information about infants of sampled mothers it was possible that we may have sampled mothers of infants who were ill or who had died. We reassured all potential participants that they would be removed from the mailing list if they did not wish to participate. A further concern was the remote possibility that there may have been mothers who were not living with their infants and who may not have wished to disclose the birth to fellow household members. Thus, we informed all potential participants (in the cover letters) that it was possible that we may have mistakenly contacted individuals who have not had a child. In this case, potential participants were instructed to contact NSO to be removed from the study either by phone or by selecting an exclusion criterion on the questionnaire itself indicating that she does not have any children.

The full study received approval from the Children's Hospital of Eastern Ontario Research Ethics Board (see appendix 8 for approval letter).

Chapter 4: Results

4.1 Results from pilot study

We contacted 30 women attending prenatal classes at the Ottawa Hospital to participate in the pilot study. A very small proportion (n=3) returned a completed questionnaire. All participants (n=3) were able to complete the study questionnaire and were all able to do so in 30 minutes or less. All three women indicated that most questions in the study questionnaire were clear and easy to understand. All participants either 'agreed' or 'strongly agreed' to the following statements about the study questionnaire:

- Questionnaire is attractive
- Questions and instructions were 'easy to follow'
- Questions were written 'in simple language that is easy to understand'

One participant indicated that the length of time to complete the study questionnaire was unreasonable. She felt as though it may be hard for new mothers to find 20 to30 minutes to complete the questionnaire and suggested that we provide the option of completing the questionnaire online. Another participant suggested that we include a 'not applicable' column to question 12 regarding potential sources of information about NBS. These suggestions were incorporated in the final version of the questionnaire.

4.2 Results from eligibility screening

Based on initial inclusion criteria (as described earlier), 25,510 mothers were identified as potentially eligible for inclusion in the study. Of these, 622 mothers were excluded because their child required repeat testing and 761 mothers were excluded because it was determined that their child had an unsatisfactory sample result. These groups would have been contacted to provide another sample and thus would be more likely to recall

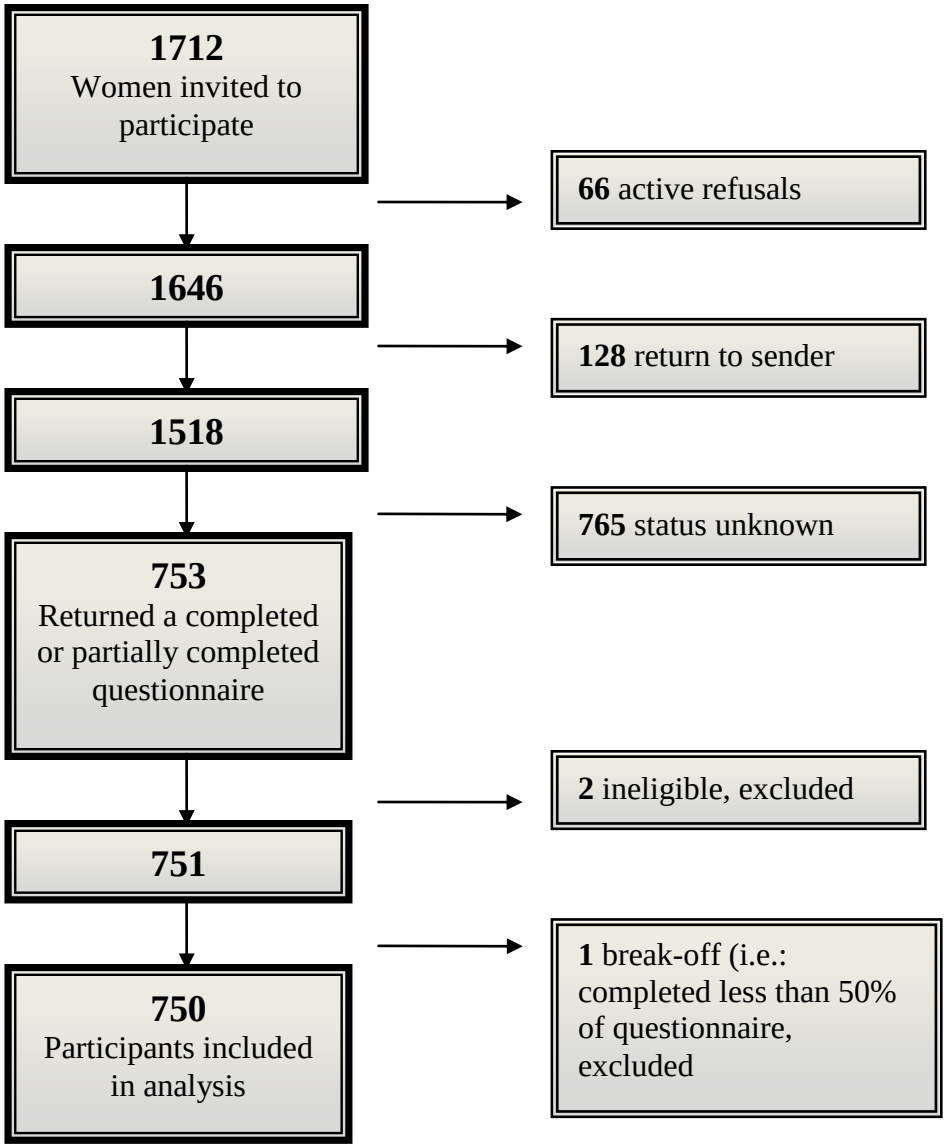
having their child tested and may have received additional education. Two mothers were excluded because their child was deceased and 231 mothers were excluded because their child received a positive screening result. We decided to exclude mothers whose infants received a positive screening result given that they would make up a small proportion of the screened population and that their experiences with NBS would likely differ from mothers whose children formed the screen negative population. Additional exclusions involved duplicate records of mothers (n=307) who had multiple children participate in the NBS program, mothers with adopted children (n=42) and mothers for whom there was insufficient contact information (n=159) (e.g., full address not in NSO database). Mothers whose youngest child was adopted were excluded since the survey included questions about receipt of prenatal information (see appendix 9, Figure 1).

4.3 Response rate

We contacted 1712 mothers to participate in the study (see Figure 2). Of these, 66 subjects actively refused to participate (either by phone or by returning a blank questionnaire) and 128 subjects were not included as a result of incorrect contact information resulting in returned mail. The status of 765 mothers is unknown (we never heard from this group, but assume that they were eligible and received our invitations to participate). The responses from 2 participants were excluded from the analyses because they met one of four exclusion criteria: one respondent was excluded because the respondent indicated that her child was older than 6 months, another was excluded because her youngest child was adopted. A third participant (break-off) was excluded from the analyses because she provided responses to less than 50% of the study questions as per a priori exclusion criteria based on standards from the American Association of Public Opinion Research for

mailed questionnaires (AAPOR).[63] Thus, reported results were based on responses from 750 mothers, for a response rate of 47% (Fig 2).

Figure 2: Disposition Status



The following formula was used to calculate the response rate in accordance with the AAPOR, response rate 2 (RR2) (AAPOR, 2009, pg. 35)[63]:

$$\textbf{RR2} = \frac{\text{complete survey + partial survey}}{\text{((Complete survey + partial survey) + (refusal + break-off+ unknown))}}$$

$$= \frac{(749 + 1)}{((749 + 1) + (66 + 1 + 765))}$$
$$= 47\%$$

In other words,

$$\textbf{Response rate} = \frac{\text{Number of usable questionnaires}}{\text{All known eligible or potentially eligible individuals in the sampling frame}}$$

(i.e.: the denominator only excludes those who we found ineligible or who we knew did not have an opportunity to participate; 2 ineligible + 128 returned to sender mail)

The majority of questionnaires received were completed in English (n=728) and were returned by mail (n=714); only 36 mothers chose to participate online.

4.4 Results from data cleaning

There were five types of problems with responses, including:

1) **Apparent incorrect selection of exclusion criteria**-where participant(s) incorrectly selected an exclusion criterion yet filled in the questionnaire
2) **Errors in following skip patterns**- where participant(s) responded incorrectly to questions involving skip patterns (ex.: questions 10-15 or questions 33-35).
3) **Unclear responses**- where participant(s) provided a response to an open-ended question which was deemed uncodeable.
4) **Multiple responses**- this occurred when participant(s) provided more than one response to a question that required a single response.
5) **Missing data**- where respondent(s) left a question (or portion of the question) blank.

For more detailed descriptions on the types of problems identified through data cleaning refer to appendix 6.

4.5 *Sample characteristics*

Most mothers in our sample were at least 25 years of age (25-29 years, 24%; 30-34 years, 40%; ≥35 years, 27%), were married or living with a partner (94%), had a college degree or higher (college, 29%; university or higher, 56%) and reported English as the language most often spoken at home (80%) (Table 5). Over 10% of the sample (n=102) reported employment as a health professional (specific employment descriptions for this group are defined in appendix 11). Of this group, 34% (n=33) reported routinely working with pregnant women or new parents (appendix 11). Just over half of the sample indicated that they had more than one child at the time of the study (53%) (Table 5). At the time of the study, the majority of mothers indicated that their youngest child was between the ages of three and less than five months (79%). Nearly all mothers reported giving birth in a hospital (97%). Obstetricians were reported most commonly as the healthcare professionals providing the most care during mothers' most recent pregnancy (61%) (Table 5).

Table 5: Sample Characteristics

	n	%	95% CI
Age (years)			
≤24	69	9	7-12
25- 29	176	24	21-27
30-34	298	40	37-45
≥35	205	27	24-31
Marital status			
Married or living with partner	701	94	92-95
Education			
High school or less	114	15	13-18
College	213	29	25-32
University or higher	409	56	52-59
Language most often spoken at home			
English	569	80	77-83
French	20	3	2-4
English and French	19	3	2-4
Other	102	14	12-17
Occupation- Health care professional	102	14	11-16
Parity			
Primiparous	352	47	43-51
Multiparous	397	53	49-57
Age of youngest child (months)			
<3	111	15	12-18
3 to <5	589	79	76-82
≥5	47	6	5- 8
Birthplace			
Hospital	729	97	96-98
Home	20	3	2-4
Primary caregiver during pregnancy			
Obstetrician	439	61	57-65
Family physician	126	18	15-21
Midwife	110	15	13-18
Nurse	33	5	3-6

Abbreviations: CI=confidence intervals

4.6 Mothers' sources of information about pregnancy, childbirth and newborn care

When asked to identify sources they used to obtain information about pregnancy, childbirth and/or newborn care, most mothers reported using books (86%) (Table 6). Other commonly reported sources included healthcare professionals (prenatal professionals, 77%; post-partum professionals, 75%), family (79%), friends (78%), and the internet (78%) (Table 6). When mothers were prompted to describe their most important source of information about pregnancy and/or childbirth, the most common responses were healthcare professionals (n=239), books (n=130) and the internet (n=79) (see appendix 10 for a complete list of responses). Similarly, when asked to identify the most important source of information about newborn care mothers commonly reported healthcare professionals (n=214), books (n=86), and family members (n=85) (see appendix 10 for a complete list of responses).

Table 6: Sources of information about pregnancy, childbirth and newborn care (Question 5)

Total N=719	**n**	**%**	**95% CI**
Prenatal class	295	41	37-45
Prenatal health professional(s)	550	77	73-80
Post-partum health professional(s)	538	75	71-78
Family members	565	79	75-82
Friends	563	78	75-81
Internet sites	562	78	75-81
Television or radio	221	31	27-34
Books	615	86	83-88

Abbreviations: CI=confidence intervals

4.7 Timing and sources of NBS education

Most respondents (89%) recalled having heard of NBS (Table 7), and could also recall their youngest child receiving NBS (93%). Fewer women could recall receiving information about NBS at any time (69%) (Table 7).

Table 7: Mothers' recollection of their experiences with newborn screening (Questions 8-10)

Total N=741	**n**	**%**	**95% CI**
Recall having heard of newborn screening			
Yes	656	89	86- 91
No	78	11	8-13
Don't know	7	1	0-2
Recall youngest child receiving newborn screening			
Yes	689	93	91-95
No	35	5	3-7
Don't know	17	2	1-4
Recall receiving information about newborn screening			
Yes	512	69	66-72
No	184	25	22-28
Don't know	45	6	4-8

Abbreviations: CI=confidence intervals

Among mothers who reported having received information about NBS, a large proportion recalled receiving this information after birth, before the heel prick (62%) and at the time of the heel prick test (72%) (Table 8). Nurses were the most commonly cited source of information about NBS (69%) followed by books (32%) (Table 8). Mothers were asked to specify any other sources of information about NBS. Of those who responded (n=26), 58% indicated that they received NBS information from print media other than books (see appendix 10). With respect to methods of communication, mothers most commonly recalled receiving information about NBS through a conversation with a healthcare professional (69%) and through the information sheet given at the time of the heel prick (68%) (Table 8).

A small number of mothers (n=4) reported other ways they recalled receiving information about NBS other than those listed in the questionnaire (see appendix 10).

Table 8: When, where and how mothers recalled receiving information about newborn screening (Questions 11-13)

	n	%	95%CI
Times at which mothers recalled receiving information [a]			
Before childbirth (during pregnancy)	245	49	45-54
After birth, before newborn screening test	309	62	58-67
At the time of the heel prick test	355	72	67-76
After the heel prick test	206	42	37-46
Sources of information about newborn screening [a]			
Prenatal class	87	19	15-23
Obstetrician	86	19	15-22
Family physician	66	14	11-18
Midwife	88	19	16-23
Pediatrician	29	6	4-9
Nurse	319	69	64-73
Family members	43	9	7-12
Friends	47	10	8-13
Internet sites	76	16	13-20
Television or radio	11	2	1-4
Books	150	32	28-37
Methods of communication about newborn screening [a]			
Conversation with a health professional	344	69	65-73
Group education	100	20	17-24
Information sheet given at the time of heel prick	340	68	64-72
Other written information	191	38	34-43
Ontario Newborn Screening Program website	23	5	3-7

[a] 230 respondents were excluded from this question because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.
Abbreviations: CI=confidence intervals

We also explored potential differences in the timing and sources of NBS education across various demographic variables (e.g., age, education, language most often spoken at home and parity). Mothers who reported speaking a language other than English at home

were significantly less likely compared to those who speak English at home to recall receiving information about NBS at any time (OR=0.46) (Table 9, bivariate results).

Table 9: Bivariate analysis: Proportion of mothers reporting that they received information about NBS by age, education, language most often spoken at home and parity (Question 10)

	Total	Reported receiving NBS information	
	n	n (%)	OR (95% CI)†
Age (years)			
≤24	67	50 (75)	Ref
25-29	172	114 (66)	0.67 (0.35-1.26)
30-34	298	210 (70)	0.81 (0.44-1.48)
≥35	204	137 (67)	0.70 (0.37-1.30)
Education			
High school or less	111	77 (69)	Ref
College	211	144 (68)	0.95 (0.58-1.56)
University or higher	407	282 (69)	1.00 (0.63-1.57)
Language most often spoken at home			
English	565	411 (73)	Ref
Other	140	77 (55)	0.46 (0.31-0.67)**
Parity			
Primiparous	349	243 (70)	Ref
Multiparous	393	270 (69)	0.96 (0.70-1.31)

* p<0.05, ** p<0.01, †unadjusted OR
Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval

Multiparous women (those with more than one child at the time of the study) were significantly less likely than primiparous women to report having received information prenatally (OR=0.65) (Table 10) (bivariate results). With regard to sources of information about NBS, multiparous women were significantly less likely than primiparous women to report receiving information from a prenatal class (OR=0.09), family members (OR=0.47), friends (OR=0.42) and from books (OR=0.63) (see appendix 11, bivariate results).

Table 10: Bivariate analysis: Proportion of mothers reporting that they received information about NBS prenatally by age, education, language most often spoken at home and parity (Question 11)

	Total	Received NBS info prenatally	
	n	n (%)	OR (95%CI)[†]
Age (years) [a]			
≤24	50	26 (52)	Ref
25-29	113	63 (56)	1.16 (0.60-2.27)
30-34	209	103 (49)	0.90 (0.48-1.66)
≥35	135	55 (41)	0.64 (0.33-1.22)
Education [a]			
High school or less	76	37 (49)	Ref
College	144	77 (53)	1.21 (0.69-2.11)
University or higher	279	127 (46)	0.88 (0.53-1.46)
Language most often spoken at home [a]			
English	409	211 (52)	Ref
Other	75	30 (40)	0.63 (0.38-1.03)
Parity [a]			
Primiparous	240	130 (54)	Ref
Multiparous	269	117 (43)	0.65 (0.46-0.92)*

* $p<0.05$, ** $p<0.01$, [†]unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval

[a] 230 respondents were excluded from this question because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

4.8 Content of NBS educational information

When asked to identify the content of educational information they had received about NBS, mothers most commonly recalled receiving information about the purpose of the NBS test (88%), how the screening test is performed (85%) and the importance of having your baby screened (72%) (Table 11). About half of mothers reported receiving information about whether they would be notified of a positive (abnormal) result (53%) or a negative (normal) result (48%). A smaller proportion (31-39%) recalled receiving information about

how they would be informed of a positive result and/or what a positive and negative result means, and 10% or fewer recalled receiving information about how results would be stored and how the blood sample would be handled after testing (Table 11).

Table 11: Information mothers recalled receiving about newborn screening (Question 14)

Total N=494[a]	**n**	**%**	**95%CI**
Information mothers recalled receiving about newborn screening			
The purpose of the newborn screening test	436	88	85-91
The importance of having your baby screened	354	72	68-76
How the screening process would be done	419	85	81-88
Whether you would be told if the result was negative (normal)	236	48	43-52
Whether you would be told if the result was positive (abnormal)	264	53	49-58
How you would be told if the result was positive (abnormal)	151	31	27-35
What a negative (normal) result means	191	39	34-43
What a positive (abnormal) result means	177	36	32-40
How the results would be stored	38	8	6-10
How the blood sample would be handled after testing	51	10	8-13

Abbreviations: CI=confidence interval

[a] 230 respondents were excluded from this question because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

4.9 Mothers' knowledge about and satisfaction with NBS

When asked to indicate their level of satisfaction with the information received about NBS there was considerable variability in responses: 19% of mothers reported being 'very satisfied', 35% reported being 'somewhat satisfied', 21% were 'neither satisfied nor dissatisfied', 18% were 'somewhat satisfied' and 7% were 'very' dissatisfied with the information they received (Table 12).

Table 12: Mothers' level of satisfaction with information received (Question 15)

Total N=506[a]	n	%	95%CI
Level of satisfaction with information received			
Very satisfied	96	19	16-23
Somewhat satisfied	178	35	31-40
Neither satisfied nor dissatisfied	107	21	18-25
Somewhat dissatisfied	91	18	15-22
Very dissatisfied	34	7	5- 9

Abbreviations: CI=confidence intervals

[a] 230 respondents were excluded from this question because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

Mothers were more likely to report being 'very' or 'somewhat' satisfied with the information they received if they recalled receiving information: prenatally (OR=2.37), from a healthcare professional (OR=5.81), or from a tear-off off sheet given at the time of the heel prick test (OR=2.21) (Table 13, bivariate results). Mothers were also more likely to report a higher level of satisfaction if they recalled receiving information about: the purpose of screening (OR=11.33), how screening would be done (OR=6.15), how results would be communicated (OR=5.53), the meaning of positive and negative results (OR=8.91), and information on the storage and handling of the sample (O=8.04) (Table 13, bivariate results).

Table 13: Bivariate analysis: Proportion of mothers who reported being 'very' or 'somewhat' satisfied with information received about newborn screening, by demographics and experience with NBS education (Question 15)

	Total	Very/somewhat satisfied with NBS info	
	n	n (%)	OR (95%CI)[†]
Age (years) [a]			
≤24	50	30 (60)	Ref
25-29	112	64 (57)	0.89 (0.45-1.75)
30-34	207	107 (52)	0.71 (0.38-1.34)
≥35	135	71 (53)	0.74 (0.38-1.43)
Education [a]			
High school or less	76	44 (58)	Ref
College	141	81 (57)	0.98 (0.56-1.73)
University or higher	279	142 (51)	0.75 (0.45-1.26)
Language most often spoken at home [a]			
English	407	220 (54)	Ref
Other	74	37 (50)	0.85 (0.52-1.40)
Parity [a]			
Primiparous	240	119 (50)	Ref
Multiparous	266	155 (58)	1.42 (1.00-2.02)
Received info prenatally [a]	246	160 (65)	2.37 (1.66-3.40)**[b]
Received info from prenatal class [a]	98	54 (55)	1.05 (0.67-1.63)[b]
Received info from HCP [a]	439	260 (59)	5.81 (3.07-10.98)**[b]
Received info from tear-off sheet [a, c]	342	206 (60)	2.21 (1.51-3.25)**[b]
Recalled receiving information about: [a]			
Purpose/benefits of screening	459	269 (59)	11.33 (4.39-29.23)**[b]
How screening would be done	432	260 (60)	6.15 (3.33-11.39)** [b]
Whether/how results are communicated	306	214 (70)	5.53 (3.74-8.19)** [b]
What negative/positive results mean	207	170 (82)	8.91 (5.79-13.70)** [b]
Policy info on storage and use	61	54 (89)	8.04 (3.58-18.06)** [b]

*p<0.05, **p<0.01, [†]unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval; HCP= Healthcare professional

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

[b] Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about information received. All of these questions were separate so responses are not mutually exclusive (e.g.: mothers could report receiving information from both pre-natal class and the tear-off sheet).

[c] The tear-off sheet is a double-sided page of information for parents that is attached to the blood collection card.

In the multivariate model final significant predictors of a high level of satisfaction with NBS education included receiving information: prenatally (OR=2.35), from a healthcare professional (OR=4.54), and from the tear-off sheet given at the time of the heel prick (OR=1.72) (Table 14). A high level of satisfaction was also significantly associated with receiving information about: the purpose and benefits of screening (OR=3.78), how results would be communicated (OR= 2.57), the meaning of positive and negative results (OR=4.19) and information about storage and handling of sample (OR= 3.13) (Table 14).

Table 14: Model 3: Predicting mothers' satisfaction (very/somewhat vs. all others) with; age, education, language, parity, whether mothers recalled receiving information about NBS prenatally, and information received

Covariates	OR (95% CI)*	*P*
Received information prenatally	2.35 (1.48-3.75)	<0.01
Received information from a HCP	4.54 (2.03-10.15)	<0.01
Received information from the tear-off sheet [a]	1.72 (1.04-2.83)	0.03
Received information about:		
Purpose/benefits of screening	3.78 (1.30-11.05)	0.02
How results will be communicated	2.57 (1.57-4.21)	<0.01
Meaning of positive/negative results	4.19 (2.52-6.99)	<0.01
Information on storage and handling of sample	3.13 (1.15-8.53)	0.03

Abbreviations: HCP= healthcare professional; OR= odds ratio; NBS= newborn screening
[a] The tear-off sheet is a double-sided page of information for parents that is attached to the blood collection card.
* Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about the source or type of information received. Odds ratios are adjusted for other variables in analysis.

Mothers were also asked a series of questions to ascertain their knowledge about NBS. Most mothers were able to correctly identify the main purpose of NBS (82%), that the sample of blood should be taken at least 24 hours after birth (74%), and that a healthcare provider would contact parents if an infant's NBS test results were positive (76%) (Table 15). However, over half of mothers responded "don't know" to a true/false question about the possibility of receiving a positive (abnormal) screening result even if an infant did not have the disease (54%) (Table 15). Further, 46% of mothers responded "don't know" and 19%

answered incorrectly to a question about whether parents have the choice not to have their infant tested (Table 15). On an exploratory basis, we developed a scoring scheme and categorized mothers into two groups (high or low knowledge) based on their responses to the knowledge questions (the number of correct responses). According to this scoring scheme, the majority of mothers had low knowledge (0-3 correct responses) about NBS (63%) (Table 16).

Table 15: Mothers' responses to knowledge questions about newborn screening (Questions 25-29)

	Correct	Incorrect	Don't know
Total N= 712	**% (95%CI)**	**% (95%CI)**	**% (95%CI)**
Main purpose of newborn screening is to identify infants who may have a disease so that treatment can begin right away to prevent serious health problems. [a]	82 (79-85)	11 (9-14)	7 (5-9)
To be most accurate, a sample of blood should be taken from the baby at least 24 hours after birth. [a]	74 (70-77)	13 (11-16)	13 (11-16)
A healthcare provider will contact you if your infant's newborn screening test results are positive. [b]	76 (72-79)	1 (0-3)	23 (20-26)
Your infant may receive a positive (abnormal) newborn screening test result even if he/she does not have the disease. [b]	35 (32-39)	11 (9-14)	54 (50-57)
You may choose not to have your infant tested. [b]	35 (31-39)	19 (16-22)	46 (42-50)

Abbreviations: CI=confidence intervals

[a] The statement shown was the correct response; a response was deemed incorrect if the respondent chose any of the distractor responses (not including if they chose 'don't know').

[b] A correct response to this true/false question was to select 'true'.

Table 16: The proportion of mothers who scored high or low on knowledge questions about newborn screening (Questions 25-29)

Total N=712	n	%	95%CI
High knowledge			
≥ 4 correct responses	264	37	34-41
Low knowledge			
≤ 3 correct responses	448	63	59-66

Abbreviations: CI=confidence intervals

Several significant bivariate predictors of knowledge were identified. Mothers were more likely to have a high level of knowledge (≥ 4 correct responses to questions about NBS) if they had a postsecondary education or higher compared to those with high school or less (college, OR= 1.79; university or higher, OR=2.43), recalled receiving any information about NBS (OR= 3.2) received information prenatally (OR= 1.58), from a healthcare professional (OR= 2.09), or from the tear-off sheet (OR= 1.98) (Table 17). Mothers were also more likely to have high knowledge if they recalled receiving information about: the purpose of screening (OR= 2.70), how screening would be done (OR= 2.57), how results would be communicated (OR=1.98), and the meaning of negative or positive results (OR=2.64) (Table 17).

Table 17: Bivariate analysis: Proportion of mothers with high knowledge (≥ 4 correct responses to knowledge questions) about newborn screening, by demographics and experience with newborn screening education. (Questions 25-29)

	Total	High knowledge about NBS	
	n	**n (%)**	**OR (95%CI)[†]**
Age (years)			
≤24	62	17 (27)	Ref
25-29	169	63 (37)	1.57 (0.83-2.98)
30-34	287	116 (40)	1.80 (0.98-3.29)
≥35	194	68 (35)	1.43 (0.76-2.69)
Education			
High school or less	104	24 (23)	Ref
College	203	71 (35)	1.79 (1.05-3.08)*
University or higher	394	166 (42)	2.43 (1.48-3.99)**
Language most often spoken at home			
English	546	213 (39)	Ref
Other	131	41 (31)	0.71 (0.47-1.07)
Parity			
Primiparous	336	117 (35)	Ref
Multiparous	376	147 (39)	1.20 (0.89-1.63)
Received NBS info	485	218 (45)	3.18 (2.19-4.61)** [b]
Received info pre-natally [a, d]	236	120 (51)	1.58 (1.10-2.27)* [b]
Received info from a HCP [a, d]	416	198 (48)	2.09 (1.19-3.65)** [b]
Received info from a pre-natal class [a, d]	89	41 (46)	1.04 (0.65-1.65) [b]
Received info from tear-off sheet [a, c, d]	334	167 (50)	1.98 (1.32-2.97)** [b]
Recalled receiving information about: [a, d]			
Purpose/benefits of screening	435	206 (47)	2.70 (1.37-5.33)** [b]
How screening would be done	411	199 (48)	2.57 (1.47-4.50)** [b]
Whether/how results are communicated	291	151 (52)	1.98 (1.36-2.88)** [b]
What negative/positive results mean	196	116 (59)	2.64 (1.81-3.84)** [b]
Info on storage and handling of sample	58	31 (53)	1.46 (0.84-2.53) [b]

*p<0.05, **p<0.01, [†]unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval; HCP= Healthcare professional

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

[b] Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about information received.

[c] The tear-off sheet is a double-sided page of information for parents that is attached to the blood collection card.

[d] All of these questions were separate so responses are not mutually exclusive (e.g.: mothers could report receiving information from both pre natal class and the tear-off sheet).

As previously indicated two multivariate models were constructed to identify potential predictors of 'high knowledge' about NBS. In the full sample of mothers, significant

predictors of having 'high knowledge' were having a postsecondary education (college, OR=1.89; university or higher, OR=2.67) and having received any information about NBS (OR=3.13) (Table 18).

Table 18: Model 1: Predicting mothers' knowledge (high vs. low) with: age, education, language, parity, and whether mothers recalled receiving information about newborn screening

Covariates	OR (95% CI)[†]	*P*
Education		<0.01
High school or less	Ref	
College	1.89 (1.06-3.34)	
University or higher	2.67 (1.57-4.53)	
Received NBS information	3.13 (2.13-4.60)*	<0.01

Abbreviations: OR= odds ratios; NBS=newborn screening

* Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about whether they could recall receiving information about screening. [†]Odds ratio are adjusted for other variables in analysis

Among women who reported having received any information about NBS at any time, predictors of having 'high knowledge' included having a postsecondary education (college, OR=1.89; university or higher, OR=2.79), reporting English as the language most often spoken at home (OR=1.96), receiving information from a tear-off sheet (OR=1.57) and receiving information about the meaning of positive and negative results (OR=2.65) (Table 19).

Table 19: Model 2: Predicting mothers' knowledge (high vs. low) with: education, language, parity, recalled receiving information about newborn screening from a tear-off sheet, and about the meaning of positive/negative results

Covariates	OR (95% CI)[†]	*P*
Education		<0.01
High school or less	Ref	
College	1.89 (0.97-3.66)	
University or higher	2.79 (1.51-5.17)	
Language		
Other	Ref	
English	1.96 (1.09-3.52)	0.03
Recalled receiving information from…		
Tear-off sheet given at the time of the heel prick test [a]	1.57 (1.01-2.43)*	0.05
Recalled receiving information about:		
Meaning of positive/negative results	2.65 (1.76-3.99)*	<0.01

Abbreviations: OR= odds ratios; NBS=newborn screening
* Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about the source or type of information received.
[a] The tear-off sheet is a double-sided page of information for parents that is attached to the blood collection card.
[†] Odds ratio are adjusted for other variables in the analysis

4.10 Mothers' opinions about NBS education

We asked mothers to indicate at what times they felt parents should receive information about NBS. The vast majority of mothers endorsed the prenatal period (during pregnancy, before childbirth) as an important time to receive information about NBS (93%) (Table 20). Over 80% of mothers also indicated that parents should receive NBS information post-natally, following birth but before the heel prick test (82%). When asked to identify the single best time for parents to receive information about NBS, over half of mothers chose the prenatal period (64%) (Table 20).

Table 20: Times at which mothers feel parents should receive information about newborn screening (Questions 16 & 17)

	Views on whether parents should receive information at these times [a]			Single best time to receive NBS information [b]		
Total N=727	**n**	**%**	**95% CI**	**n**	**%**	**95% CI**
During pregnancy, before childbirth	676	93	91-95	467	64	61-68
After birth, before heel prick test	594	82	79-84	198	27	24- 31
At the time of the heel prick test	514	71	67-74	56	8	6-10
After the heel prick test	370	51	47-55	<5	--[d]	--[d]
No opinion	n/a [c]	n/a [c]	n/a [c]	<5	--[d]	--[d]

Abbreviations: CI=confidence intervals

[a] Proportion of respondents agreeing (participants rated each item separately)

[b] Respondents were asked to select only one item indicating when they feel is the single best time for parents to receive information about Newborn Screening.

[c] n/a- This item was not applicable to the question.

[d] -- Proportions not shown because frequency counts were less than five.

Obstetricians were selected most often as the health professional expected to be most knowledgeable about NBS (32%) followed by pediatricians (22%) and nurses (19%) (Table 21). A small number of mothers indicated another health professional (not listed) who they expected to be most knowledgeable about NBS (n=8) (appendix 10).

Table 21: Healthcare professional who mothers expect to be the most knowledgeable about newborn screening (Question 18)[a]

Total N=698	**n**	**%**	**95%CI**
Obstetrician	220	32	28-35
Family physician	91	13	11-16
Midwife	52	7	6-10
Pediatrician	155	22	19-25
Nurse	136	19	17-23
No opinion	36	5	4-7

Abbreviations: CI=confidence intervals

[a] Respondents were asked to select only one item indicating which healthcare professional they expect to be the most knowledgeable about newborn screening.

Mothers were asked to identify important ways to receive information about NBS. An overwhelming majority selected 'conversation with a healthcare professional' (95%) and a high proportion also endorsed 'information sheet given at the time of the heel prick' (84%)

(Table 22). When asked to specify the single best way to receive information about NBS more than half of mothers chose conversation with a healthcare professional (64%) (Table 22). Few mothers (n<5) provided other important ways to receive information (appendix 10).

Table 22: Important ways, according to mothers, for parents to receive information about newborn screening (Questions 21 & 22)

	Views on how parents should receive NBS information [a]			Single best way to receive NBS information [b]		
Total N=716	**n**	**%**	**95% CI**	**n**	**%**	**95% CI**
Conversation with a HCP	680	95	93-96	457	64	60-67
Group education (ex.: prenatal class)	550	77	74-80	35	5	3-7
Information sheet	601	84	81-87	113	16	13-19
Other written information	567	79	76-82	91	13	10-15
Ontario newborn screening website	528	74	70-77	20	3	2-4

Abbreviations: CI=confidence intervals

[a] Proportion of respondents agreeing (participants rated each item separately)

[b] Respondents were asked to select only one item indicating the single best way for parents to receive information about newborn screening.

When asked to specify the importance of including various topics in education to inform parents about NBS, most of the topics were rated as 'very important' by over 70% of mothers (Table 23). The most highly endorsed topics in terms of the proportion of mothers rating a topic as 'very' important were the purpose of the NBS test (97%), whether parents will be told about positive (abnormal) results (95%), what a positive (abnormal) result means (94%) and the importance of the NBS test (91%) (Table 23).

Table 23: Information mothers feel is very important to include in education about newborn screening to parents (Question 23)
Abbreviations: CI=confidence intervals

Total N= 733	n	%	95%CI
The purpose of the newborn screening test	711	97	95-98
Whether you would be told if the result was positive (abnormal)	696	95	93-96
What a positive (abnormal) result means	691	94	92-96
The importance of having your baby screened	664	91	88-93
Whether you would be told if the result was negative (normal)	625	85	82-88
What a negative (normal) result means	595	81	78-84
How you would be told if the result was positive (abnormal)	585	80	77-83
How the screening process would be done	540	74	70-77
How the results would be stored	293	40	36-44
How the blood sample would be handled after testing	269	37	33-40

Several participants (n=127) identified additional information that they believe parents should be told about NBS. The most common responses were that parents should be told about the disorders included in the screening program (n=24), and what to do after receiving a positive result (n=17) (appendix 10).

Chapter 5: Discussion

5.1 Evidence for study design

We invited 1712 mothers to participate in a self-administered (mailed) survey about NBS education. We chose this method given the growing evidence which supports the benefits of using self-administered questionnaires over traditional telephone methods for studies of the general population, for example due to emerging challenges such as increased use of call screening and blocking technologies.[62,64] There is also evidence in the literature to show that multiple contacts are an effective strategy for improving response rates for mailed surveys,[62,65] and so in an attempt to reduce non-response bias we decided to contact mothers up to five times (following Dillman's tailored design method for mailed surveys).[62]

5.2 Mothers' awareness of NBS

When asked about their experiences with NBS, most mothers participating in our study reported having heard of NBS (89%) and could recall their youngest child receiving NBS (93%). While similar findings were reported by Davey et al (93% of mothers reported having heard of NBS),[49] other studies have reported much lower proportions of parents being aware of NBS.[43,48,52] It is possible that the apparently high level of awareness about NBS among mothers in our sample may in part be due to mothers confusing NBS with prenatal screening or with newborn hearing screening. In support of this hypothesis, 41 women in our sample indicated that their child received a positive NBS test result, while NSO confirmed that all infants of mothers in the sampling frame had received negative screening results. Another likely explanation for the high awareness about NBS is that the true population proportion of mothers who are aware of NBS was overestimated in our study

due to non-response bias(this is further discussed in the limitations section). Finally, it is also possible that this finding is an accurate estimate for this population (i.e., awareness about NBS may be relatively high among mothers of young infants in Ontario).

5.3 Mothers' recollection and the importance of NBS education

Despite this apparently high level of awareness of NBS, fewer than 70% of participants reported actually receiving any information about NBS at any time. To understand the potential consequences of this apparent lack of communication for a substantial minority of parents, one must first consider the purposes of pre-screening education. As previously mentioned, pre-screening education may be important to maximize the benefits of NBS (educating parents about the purpose, benefits, and process of screening may improve follow-up rates for positive results)[37] and to mitigate potential psychosocial harms (educating parents about the possibility of receiving either a true or false positive result may help to prepare them for such results).[23,35,36] To achieve these goals, it follows that parents need to be well-informed about the purpose, benefits, and process of NBS and about the meaning of different types of results (i.e., there is a need for high knowledge of these aspects of NBS).

Aside from follow-up of positive results and preventing psychosocial harm, are there other reasons we should be concerned if some parents are receiving little or no information about NBS? To date most Canadian and US NBS programs operate on an implied consent basis (although this practice has been challenged by some in the context of expanded screening as mentioned earlier).[21,22] If we assume that this implied consent model is likely to continue, one may question whether parents need any education beyond that which is

necessary to achieve appropriate follow-up and mitigation of harm. However, several studies have noted that parents themselves consider NBS education important.[43,49] The notion that parents have an expectation to be provided with education about NBS supports the assertion by Manson that there are several reasons why patients may want information about interventions independent of their desire to take part in decision making about those interventions, such as: 1) to make decisions about things other than treatment options (for example who will take the infant to hospital if further testing is needed), 2) to be psychosocially and emotionally prepared (as already discussed), 3) to understand the reasons for different treatment options, 4) to feel respected (failure to provide information may be indicative of lack of respect), 5) to ensure trustworthiness (provision of information may facilitate confidence in the healthcare provider or in this case the screening program), or 6) to have the opportunity to interact (ask questions, seek clarification).[66]

Thus, effective NBS education practices may be important because parents need knowledge to achieve the benefits and minimize the harms of screening; and because parents expect and want education and this contributes to their satisfaction with and trust in the screening program. In addition to ensuring that all mothers receive education about NBS, screening programs also need to consider how best to provide education (when, who, in what format) and what content this education should include. If the purpose of education is both to achieve outcomes that require understanding of specific aspects of NBS and to improve parental experiences and foster trust, then both parental knowledge and parental satisfaction are appropriate metrics for evaluating the effectiveness of educational strategies.

Our study examined the different dimensions of NBS education (timing, sources, methods, and content). Within each of these dimensions, we investigated mothers'

experiences with NBS education, their opinions about the ways in which education should be delivered, and the characteristics of their experiences that were most strongly related to knowledge and satisfaction.

5.4 Timing of education

Among mothers who could recall receiving any information about NBS, a higher proportion reported receiving this during the post-natal period (after birth before the screening test, 62%; at the time of the heel prick test, 72%), rather than prenatally (49%). This concurs with the findings from other studies which report the postnatal period as the most common time for parents to receive NBS information.[4,21,43,47,49,51,53,54] This current practice of providing information postnatally is contrary both to recommendations published in the US and UK guidelines[6,44] and to previous studies of parental views on when NBS information should be provided.[43,49] Mothers in our sample agreed with participants in those previous studies, with 93% endorsing the prenatal period as a time when parents should receive information. More than half of participants felt that this was the *single* best time for parents to receive information (64%). Parental preference for receipt of information prenatally makes sense given that the post-natal period is often a time when parents are pre-occupied with caring for their newborn and may be overwhelmed with other information about newborn care. An important question to consider is how timing of education affects knowledge or understanding about and satisfaction with NBS education. Our study showed that receipt of information prenatally was associated with higher satisfaction (OR=2.35) compared with receiving information postnatally only, but not with higher knowledge about NBS.

5.5 Sources and method of education

If pre-screening education about NBS is to be provided prenatally, identifying where parents currently obtain prenatal information about pregnancy, childbirth or newborn care may help to target education strategies. Mothers in our sample most commonly reported receiving this information from books (86%), family members (79%), internet sites (78%), friends (78%) and prenatal health professionals (77%). The most important source of information about pregnancy and childbirth according to mothers was health professionals (n=239). To be effective, education strategies should target these popular sources.

Our study findings, specific to NBS education, further highlight that targeting health care professionals is a crucial education strategy for the provision of information, particularly prenatally. We asked mothers to identify where and how they received information about NBS. Over half of mothers (69%) indicated that they received this information from nurses, with books a distant second choice (32%). The most common methods of receiving information were through conversation with a healthcare provider (69%) and from an information sheet given at the time of the heel prick (68%). Receiving information through a conversation with a healthcare provider was consistent with participants' views about how they wanted to receive information: 95% of mothers felt parents should receive information through conversation with a health care provider and 64% of mothers indicated that this was the single best way for parents to receive information about NBS. Supporting these findings, receiving information from a healthcare provider was associated with higher satisfaction (OR 4.54), although not with higher knowledge. However, consistent with our finding that mothers desired but were not receiving education during the prenatal period, a higher proportion of mothers expected obstetricians to be the

most knowledgeable health care providers about NBS (32%) compared with pediatricians (22%) and nurses (19%); yet only one fifth of mothers actually reported receiving information from obstetricians.

These findings concur with other studies which examine providers' practices and attitudes towards NBS education. According to Faulkner et al, less than half of prenatal care providers report discussing NBS with their patients and they had mixed perceptions about who was responsible for providing this information.[53] Similarly, in a recent study of Ontario health care professionals, midwives and nurses were most likely to report discussing NBS with parents (98% and 47% respectively) while obstetricians, family physicians and pediatricians were least likely (<20%) to report doing this.[21] In that study providers who perceived a responsibility to inform parents were 3 times more likely to report doing so compared to those who did not perceive this responsibility. Those who lacked confidence in their ability to educate parents were 70% less likely to engage in discussions about NBS with parents.[21]Further, physicians were more likely than non-physicians (midwives and nurses) to report lack of time and compensation as barriers to discussing NBS with parents.[21] Obstetricians were least likely to perceive a responsibility of informing parents and were most likely to cite a lack of time as a barrier (65%).[21] According to the Provincial Perinatal Surveillance System Committee, obstetricians are the most common prenatal primary caregivers in Ontario,[67] suggesting that if mothers are to receive prenatal information about NBS through a conversation with a health care provider (as they seem to prefer), this professional group will need to be involved.

Significantly, the ACOG recently released a report endorsing the prenatal period as an ideal time for parents to receive information and specifically suggested providing parents

with NBS information in the first trimester at the first obstetric visit along with other patient information.[22] It will be interesting to follow the response of obstetricians to this guideline from their US professional college. There may be a need to address barriers such as confidence, perceived lack of time and compensation through targeted provider education.

A conversation with a health care provider was not the sole important method of receiving NBS education in our study. Indeed, receiving information from the tear-off sheet given at the time of the heel prick was predictive of both high knowledge (OR=1.57) and higher satisfaction (OR=1.72) among participants. These findings support the suggestion to provide multiple, brief, clear explanations about NBS at several time points and in different ways. [47]

5.6 Content of NBS education

A high proportion of mothers in our study recalled receiving information about the purpose (85-91%) and importance of screening (68-76%) and indeed the majority felt this information was important for parents to receive (purpose, 95-98%; importance, 88-93%). This concurs with recent US guidelines about the content of NBS education (Davis et al, 2006; AAP, 2000). This also agrees with previous studies that found that written educational materials have tended to emphasize the purpose and benefits of NBS more than other elements, such as the potential harms.[45,56-58] Study participants were more likely to report higher satisfaction if they recalled receiving information about the purpose of screening (OR=3.78) but this was not independently associated with higher knowledge.

Fewer mothers recalled receiving information about the process of communication of NBS results (<60%) and about the meaning of positive and negative results (<40%); and

only 35% correctly answered the knowledge question that identified the possibility of a false positive result. Most participants (80%) felt this information was important. Being informed about the communication of results was also associated with higher satisfaction in our sample (OR=2.57) and receiving information about the meaning of positive and negative results was a predictor of both higher knowledge (OR=2.65) and higher satisfaction (OR=4.19). The views of mothers in our study and the association of this information with knowledge and satisfaction support the AAP guidelines, which state that parents should be informed about the possibility of receiving a false-positive result.[6] Interestingly, this was not included in the guidelines developed by Davis et al, which were informed by focus groups with parents.[43] This exclusion from these recent guidelines conflicts with the concern that this information may be important to prevent psychosocial harm, as discussed earlier.

Also included in the AAP guidelines but not in those developed by Davis et al is information about program policies for sample storage and secondary use.[6] A very small proportion of mothers in our study recalled receiving information about the storage and handling of samples (10% or less) and few mothers found this information to be of great importance (<50%) relative to other informational messages. However, this apparently lower proportion who viewed this information as essential or very important may have resulted from the way in which the question was worded in our questionnaire. It is possible that mothers were unaware that blood samples are retained and that they may be used for subsequent research (informing parents about this may help impress upon them the importance of such subsequent research). Perhaps more mothers would consider this information to be important if the question were worded differently, for example; "How important is it for parents to receive information about how long samples are stored or

whether and how they will be used after testing (for example, for research)?" Ethical and legal issues related to policies about the storage and secondary use of samples have received considerable attention in the literature in recent years [68-71] and parents may become increasingly aware of the associated privacy concerns. Although this information may not have been prioritized by participants in our study, if such privacy concerns do become more prevalent, the provision of parental pre-screening education about such policies may be an important way to protect public trust in the screening program. Supporting this, receiving information about the storage and handling of the samples was independently associated with higher satisfaction in our study (OR=3.13).

5.7 Limitations

The most important limitation of this study was the potential for non-response bias. The response rate of 47% is consistent with other surveys of similar population samples,[49,60] but low enough to be concerned about potential differences between responders and non-responders. No comprehensive data are available to define the demographic distribution of new mothers in Ontario. Thus, to evaluate the representativeness of our sample, we made comparisons to the demographic characteristics reported in the national 2008 Canadian Maternity Experiences Survey (CMES). Relative to participants in that study we found that a higher proportion of mothers in our sample were between the ages of 30-34 years (40% vs. 33%), had a university level education or higher (56% vs. 34%), and were married or living with a partner (94% vs. 90%) (appendix 14).[61] Similar characteristics were observed across the two studies with respect to parity (multiparous 53% vs. 55%; primiparous 47% vs. 45%) (appendix 14).[61] Although the CMES was national rather than Ontario-based and may itself have been characterized by non-response bias these findings suggest that there may be

concerns about the representativeness of our sample with respect to age and education. This is potentially important given that education was significantly associated with some NBS experience variables (e.g., sources of NBS information) and was associated with having high knowledge about NBS. Not surprisingly, speaking English at home was also a predictor of high knowledge about NBS. We likely have an overrepresentation of English speakers in our sample, since a high degree of skill in comprehending either English or French was required to complete the 10-page questionnaire.

A further limitation of the study was the construction of new questions, not previously validated, to represent the constructs of interest. For example, there are no pre-existing validated measures of knowledge about NBS. Our findings with respect to the knowledge score were logically coherent (e.g., predictors of higher knowledge included higher education, speaking English at home, and reporting having received information about NBS, which would be expected), which supports the validity of our questions. However, further validation work would be useful in evaluating and refining the measures.

5.8 *Future research direction*s

The findings from this study generated some potential hypotheses that may be explored further in future studies, for example:

1. Provision of information prenatally will be associated with higher knowledge and satisfaction
2. Providing information about the meaning of results will be associated with higher knowledge and satisfaction.

Additional research questions that have become apparent as a result of this study that may be further investigated include:

- What are the barriers to and facilitators of delivering pre-screening education about NBS among parental healthcare providers?
- What are the most appropriate outcomes of NBS education (and do these differ from different perspectives, for example, among public health policy decision-makers as compared with parents)?
- What constitutes high knowledge about NBS among parents, and how might this best be measured?

5.9 *Future modification of the study questionnaire*

As previously mentioned there were no pre-existing validated measures with which we could represent the constructs of interest and as a result the questionnaire was comprised of novel questions. There were a small number of questions, as discussed earlier (mainly those that involved multiple items in a series, e.g., question 12) that appeared to be problematic for many responders (specifically, participants tended to treat these items as "check-all-that-apply" rather than answering each individually). Although we explicitly avoided using a "check-all-that-apply" format so as to encourage thoughtful responses, in hindsight, these questions may be improved by using a response scale (similar to question 23, see appendix 4) that asks for a rating beyond "yes" or "no".

As described, the knowledge questions also require further validation. A current debate about knowledge questions in survey research centres on the value of including a "don't know" response option for true-false or multiple choice questions. We chose to

allow participants to select "don't know". It is possible that constructing questions this way may have led to an underestimation of knowledge (some participants may have selected don't know even if they may have truly been able to answer the question). In our study, this is likely balanced by a potential overestimation of knowledge that may have resulted from non-response bias (those who chose not to respond may have been less knowledgeable about NBS compared to respondents). Importantly, for the purposes of estimating knowledge about NBS, it seemed appropriate to be able to distinguish between parents who were uninformed and those who may have been misinformed. This is an important methodological question that would benefit from further discussion in future use of the questionnaire or in the further development or refinement of measures of parental knowledge about NBS.

This questionnaire may be useful for future studies that may wish to evaluate mothers' experiences, knowledge and opinions about NBS, in Canadian jurisdications or internationally. The questionnaire could be modified to better suit the needs of investigators working in the context of different NBS programs (for example, the "tear-off sheet" is a method of education used specifically in Ontario). Parts of the questionnaire may also be useful for evaluating educational interventions (for example, to compare the impact of receipt of information prenatally vs postnatally in terms of the impact on parental knowledge and satisfaction).

Chapter 6: Conclusion

Effective pre-screening education is an important component of the NBS system. Informing parents about the purpose, benefits, process, and possible results of screening may facilitate prompt follow-up following an initial positive result and may mitigate potential psychosocial harm. Effective education may also be a valuable tool to enhance parental trust in the NBS program and to promote a positive experience with NBS. The results from this study show that while 93% of mothers recalled their youngest child receiving screening, fewer than 70% actually recalled receiving any information about NBS. In addition to the need to ensure that all parents receive education about NBS our study identified several specific priorities for future educational initiatives regarding NBS in Ontario:

- Consistent with previous research and with available guidelines, mothers in our sample demonstrated a strong preference for receiving information prenatally. However, fewer than half the participants who reported receiving any information recalled receiving it during this time. Receipt of information prenatally was a strong predictor of higher reported satisfaction, further emphasizing its importance in fostering positive experiences with the screening program.
- Mothers in this study identified obstetricians as the healthcare professional they expected to be the most knowledgeable about NBS and the vast majority received most of their prenatal care from an obstetrician. However, fewer than 20% of participants who received NBS education reported receiving information from obstetricians. Given mothers' preferences for the prenatal period, the majority opinion that a conversation with a healthcare provider is the most important way to receive NBS education, and the finding that receipt of information from a healthcare provider was associated with higher

satisfaction, the results point to a clear need to engage prenatal healthcare providers, particularly obstetricians, about the importance of discussing NBS with parents. Since receiving information from a tear-off sheet given at the time of the heel prick was predictive of both higher knowledge and higher satisfaction, our results also support the practice of providing education at multiple time points and through multiple sources.

- Mothers appeared well-informed about the purpose, benefits, and process of screening, based on the messages they recalled receiving and their responses to the knowledge questions. However, they were less knowledgeable about the possibility of a false positive result and fewer than 40% of those who received information reported learning about the meaning of positive and negative results. Receipt of information about the meaning of results was a predictor of both high knowledge and high satisfaction; and one of the proposed benefits of NBS education is to prepare parents for different types of results. Therefore, we suggest targeting this aspect of NBS as a content priority in parental pre-screening education.

By linking the purposes of effective parental pre-screening education to the outcomes of knowledge and satisfaction, and by considering mothers' experiences and their opinions in the context of when, who/how, and what education is or could be provided, this study enabled us to identify strengths and challenges in NBS education in Ontario. The findings also have relevance for other jurisdictions with expanded NBS programs.

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Appendix 1: List of disorders currently screened by Newborn Screening Ontario

Table 24: Conditions included in Ontario screening panel[13]

- Argininosuccinic Acidemia (ASA)
- Citrullinemia
- Homocystinuria
- Maple Syrup Urine Disease (MSUD)
- Phenylketonuria
- Tyrosinemia Type 1
- B-Ketothiolase (BKT) Deficiency
- Glutaric Acidemia Type 1 (GAI)
- 3-Hydroxy-3-methylglutaryl CoA Lyase Deficiency
- Isovaleric Acidemia (IVA)
- 3-Methylcrotonyl-CoA Carboxylase Deficiency
- Methylmalonic Acidemia (mutase deficiency and cobalamin defects)
- Multiple Carboxylase Deficiency
- Proprionic Acidemia
- Carnitine Uptake Disorder (CUD)
- LCAD Deficiency
- MCAD Deficiency
- Trifunctional Protein Deficiency
- VLCAD Deficiency
- Congenital Adrenal Hyperplasia (CAH)
- Congenital Hypothyroidism (CH)
- Cystic Fibrosis (CF)
- Biotinidase Deficiency
- Galactosemia
- Sickle cell anemia
- Hb SC Disease
- Sickle/Thalassemia

Appendix 2: Sample size calculations

The following formula was used to determine the sample size needed for this study:

$$n = p(1-p)\frac{[Z_{1-(\alpha/2)}]^2}{E}$$

$$n = 0.25\frac{[Z_{1-(\alpha/2)}]^2}{0.05}$$

$$n = 0.25\frac{[1.96]^2}{0.05}$$

$$n = 384.2$$

Therefore, assuming a 50% response rate, the total number of subjects required is:
n= 385 x 2
n=770

The sample size was further inflated to account for the fact that at least 45% of the sample will be primiparous (have one child).

n=770/0.45
n=1712

Therefore the final sample size for this survey was 1712.

Appendix 3: Pre-notification letter, contact letters, participant information form and post-card

[date]

[name of potential participant] [ID number]
[address of potential participant]

Dear [name of potential participant],

Newborn screening (NBS) is a program funded by the government of Ontario. The goal of the newborn screening program is to identify infants at-risk for certain rare but serious diseases. Almost all babies in Ontario are screened for at least 28 of these diseases and there are many ways we try to make sure that parents are informed of newborn screening. Your baby was screened using a heel prick blood sample taken in the first few days of life, and you should have received information about the program around the time of your child's birth.

I am writing you as the Director of the Ontario Newborn Screening Program to invite you to participate in a research study about newborn screening education.

We are interested in how new mothers learn about newborn screening during or after pregnancy. We are also seeking mothers' opinions about how best to educate parents about newborn screening. We hope to use the results of this research to improve the education offered to new mothers in the future.

The research study has been approved by the Children's Hospital of Eastern Ontario (CHEO) Research Ethics Board. The study is being run by the Ontario Newborn Screening Program (Christina Honeywell and Dr. Pranesh Chakraborty) together with researchers from the University of Ottawa (Makda Araia, Dr. Beth Potter, and Dr. Brenda Wilson).

Staff of the Ontario Newborn Screening Program randomly selected mothers with infants aged 6 months or younger who had normal screening results in early 2009. These mothers are being contacted about participating in this research study. We do not have any health information about your child beyond the screening result, and so we may have contacted mothers whose infants are ill or deceased. It is also possible that we have mistakenly contacted women who have not had a child recently. **If we have contacted you in error we apologize and ask that you notify us so we can remove your name from the mailing list.** This list of mothers is kept securely by the Ontario Newborn Screening Program. **At no point during the study will researchers outside the screening program be given your name or address unless you wish it**.

The study uses a questionnaire, which you will be receiving in the mail within the next week. Included with the questionnaire is a Participant Information Form that gives a more

detailed description of the project. Please read the Participant Information Form before deciding whether or not to take part. You will also receive a small gift with the questionnaire as a way of saying thanks.
The researchers we are working with at the University of Ottawa will have access to the questionnaires from this project. The questionnaires will not include any names or other identifying information. We would like to reassure you that no access to any information whatsoever on your child (even whether you have a son or daughter) will be given to anyone outside the Ontario Newborn Screening Program. If you decide that you would like to participate, fill in the questionnaire when it arrives and return it to the Ontario Newborn Screening Program in the postage-paid envelope provided.

We understand that you may prefer not to take part in this study. If this is the case you may do one of the following things:

1. Return a blank questionnaire in the postage-paid envelope provided – we will immediately take your name off the mailing list for this study.

2. Call the Ontario Newborn Screening Program [xxx xxx-xxxx ext. xxxx] and leave a voicemail message – we will immediately take your name off the mailing list for this study.

3. Ignore this letter. The newborn screening program will contact you up to 4 more times to invite you to participate in the study. This is done to try to get as many responses as possible which will help researchers be more certain of the results. If you ignore this and all four remaining contacts your name will be taken off the mailing list for this study.

Whatever you decide to do, you are under no obligation to the Children's Hospital of Eastern Ontario, the Ontario Newborn Screening Program, or the University of Ottawa. Your choice to participate or not in this project will not affect any future care you may receive at CHEO. You are free to withdraw from this study at any time and there will be no penalty to you or your child. I appreciate the time you have taken to read this letter.

Yours truly,

Dr. Pranesh Chakraborty
Laboratory Director/Ontario Newborn Screening Program
Children's Hospital of Eastern Ontario

(Valid until July 6, 2010)

Contact letter #2

[date]

[name of potential participant] [ID number]
[address of potential participant]

Dear [name of potential participant],

You may remember receiving a letter a few days ago regarding a research study about newborn screening. I am writing to you today to invite you to participate in this study. We are interested in how mothers learn about newborn screening during and after pregnancy. We are also interested in mothers' opinions about how best to educate parents about newborn screening.

The study has been approved by the Children's Hospital of Eastern Ontario (CHEO) Research Ethics Board. It is being run by the Ontario Newborn Screening Program (Christina Honeywell and Dr. Pranesh Chakraborty) together with researchers from the University of Ottawa (Makda Araia, Dr. Beth Potter, and Dr. Brenda Wilson).

You were chosen by staff of the Ontario Newborn Screening Program from a list of names of mothers with infants aged 6 months or younger who had normal screening results in early 2009. I can confirm that your name and contact information will never be seen by anyone outside the newborn screening program.

This study uses a questionnaire. Included with this letter are the questionnaire and a Participant Information Form. The Participant Information Form gives a more detailed explanation of the study. Please read the form before deciding whether or not to take part in the study. We have also included a small gift (a $2 gift certificate for a coffee shop) as a way of saying thanks (please keep this whether or not you decide to participate).

If you decide that you would like to take part, please fill in the questionnaire and return it to the Ontario Newborn Screening Program in the postage-paid envelope provided. Please keep the Participant Information Form.
You may also participate in this study by filling in the questionnaire online. To do this, please type in the website address provided below. Then enter your username and password when prompted. This will allow you to securely access the questionnaire. It will also allow you to return to where you left off if you are not able to complete the entire questionnaire in one session.

Website address: <insert URL>
Your username:
Your password:

We understand that you may prefer not to take part in this study. If this is the case, please refer to the Participant Information Form for instructions on how to have your name removed from the mailing list for this study.

Even if you decline to take part, please keep the Participant Information Form.

Taking part in research is voluntary. Whatever you decide to do, you are under no obligation to the Children's Hospital of Eastern Ontario, the Ontario Newborn Screening Program, or the University of Ottawa.

Your choice to participate or not in this project will not affect any future care your child may receive at CHEO. You are free to withdraw from this study at any time and there will be no penalty to you or your child. I appreciate the time you have taken to read this letter.

Sincerely,

Dr. Pranesh Chakraborty
Laboratory Director/Ontario Newborn Screening Program
Children's Hospital of Eastern Ontario

(Valid until July 6, 2010)

Contact letter #4

[date]

[name of potential participant] [ID number]
[address of potential participant]

Dear [name of potential participant],

I am writing to you as a follow-up to remind you that you are invited to participate in a research study about newborn screening. You may remember that the purpose of this study is to understand how mothers learn about newborn screening during or after pregnancy. We are also interested in their opinions on how best to educate parents about newborn screening.

The study has been approved by the Children's Hospital of Eastern Ontario (CHEO) Research Ethics Board and is being run by the Ontario Newborn Screening Program (Christina Honeywell and Dr. Pranesh Chakraborty) together with researchers from the University of Ottawa (Makda Araia, Dr. Beth Potter, and Dr. Brenda Wilson).

Staff of the Ontario Newborn Screening Program randomly selected mothers with infants aged 6 months or younger who had normal screening results. No information about you or your child will be given to anyone outside the Ontario Newborn Screening Program.

This study uses a questionnaire. Included with this letter are the questionnaire and a Participant Information Form. The Participant Information Form gives a more detailed explanation of the study. Please read the Form before deciding whether or not to take part in the study.

If you decide that you would like to take part, please fill in the questionnaire and return it to the Ontario Newborn Screening Program in the postage-paid envelope provided. Please keep the Participant Information Form.

You may also participate in this study by filling in the questionnaire online. To do this, please type in the website address provided below. Then enter your username and password when prompted. This will allow you to securely access the questionnaire. It will also allow you to return to where you left off if you are not able to complete the entire questionnaire in one session.

Website address: <insert URL>
Your username:
Your password:

We understand that you may prefer not to take part in this study. If this is the case, please refer to the Participant Information Form for instructions on how to have your name removed from the mailing list for this study.

Even if you decide not to take part, please keep the Participant Information Form.

Please remember that whatever you decide you are under no obligation to the Children's Hospital of Eastern Ontario, the Ontario Newborn Screening program, or the University of Ottawa.

Your choice to participate or not in this project will not affect any future care your child may receive at CHEO. You are free to withdraw from this study at any time with no penalty to you or your child. I appreciate the time you have taken to read this letter.

Sincerely,

Dr. Pranesh Chakraborty
Laboratory Director/ Ontario Newborn Screening Program
Children's Hospital of Eastern Ontario

(Valid until July 6, 2010)

Contact letter #5

[date]

[name of potential participant] [ID number]
[address of potential participant]

Dear [name of potential participant],

This is a final letter to invite you to participate in a research study about newborn screening.

As you may remember, we are interested in how mothers learn about newborn screening during or after pregnancy. We are also interested in mothers' opinions on how best to educate parents about newborn screening.

The study has been approved by the Children's Hospital of Eastern Ontario (CHEO) Research Ethics Board. It is being run by the Ontario Newborn Screening Program (Christina Honeywell and Dr. Pranesh Chakraborty) together with researchers from the University of Ottawa (Makda Araia, Dr. Beth Potter, and Dr. Brenda Wilson).

Staff from the Ontario Newborn Screening Program randomly selected mothers of infants aged 6 months or younger who received normal screening results. No information about you or details of your child will be given to anyone outside the newborn screening program.

This study uses a questionnaire. Included with this letter are the questionnaire and a Participant Information Form. The Participant Information Form gives a more detailed explanation of the study. Please read the form before deciding whether or not to take part in the study. To participate, please fill in the questionnaire and return it to the Ontario Newborn Screening Program in the postage-paid envelope provided. Please keep the Participant Information Form.

You may also participate in this study by filling in the questionnaire online. To do this, please type in the website address provided below. Then enter your username and password when prompted. This will allow you to securely access the survey. It will also allow you to return to where you left off if you are not able to complete the entire questionnaire in one session.

Website address: <insert URL>
Your username:
Your password:

If you have decided not to take part, we would appreciate it if you would return the blank questionnaire in the postage-paid envelope provided. You may also choose to simply ignore

this letter (and throw out the questionnaire). If we do not hear from you, we will take your name off the mailing list for this study and you can expect no further contact from us. I thank you for taking the time to read this letter, and I wish you and your family all the best for the future. I suggest that you keep the Participant Information Form for future reference.

Please remember that whatever you decide you are under no obligation to the Children's Hospital of Eastern Ontario, the Ontario Newborn Screening Program, or the University of Ottawa.

Your choice to participate or not in this study will not affect any future care your child may receive at CHEO. You are free to withdraw from this study at any time with no penalty to you or your child. I appreciate the time you have taken to read this letter.

Sincerely,

Dr. Pranesh Chakraborty
Laboratory Director/Ontario Newborn Screening Program
Children's Hospital of Eastern Ontario

(Valid until July 6, 2010)

Mothers' Views and Experiences with Newborn Screening

Investigators: Dr. Pranesh Chakraborty, Christina Honeywell (Ontario Newborn Screening Program),
Dr. Beth Potter, Makda Araia and Dr. Brenda Wilson (University of Ottawa)

Questionnaire Survey

PARTICIPANT INFORMATION FORM

You are invited to take part in a research study. This Participation Information Form describes the study. It outlines what will be asked of you if you decide to take part.

Purpose of this Research Study:

We are inviting you to take part in this study because we believe that mothers of young infants are likely to have received information about newborn screening. This is because almost all babies in Ontario receive newborn screening. We are interested in how new mothers learn about newborn screening. We are also interested in mothers' opinions about how best to educate parents about newborn screening. We hope to use the results of this research to improve the education offered to new mothers in the future.

Participants:

This study will include about 1700 mothers whose infants have recently had newborn screening. We are inviting mothers whose infants received normal newborn screening results. However, we do not have any health information about your child beyond the screening result. As a result, we may have contacted mothers whose infants are ill or deceased. It is also possible that we have mistakenly contacted women who have not had a child recently. **If we have contacted you in error we apologize and ask that you notify us so we can remove your name from the mailing list.**

Procedures:

This study uses a questionnaire. The questionnaire is included with this package and should take about 30 minutes to complete. If you would like to participate, please fill in either the English or French questionnaire by yourself. Then please mail it back to the Ontario Newborn Screening Program, using the postage-paid envelope provided. The study overall is expected to take about six months.

Instead, you may choose to participate in this study by filling in the questionnaire online. Instructions for completing the questionnaire online are provided in the letter that accompanies this form.

We understand that you may prefer not to take part in this study. If this is the case you may do one of the following things:

1. Return a blank questionnaire in the postage-paid envelope provided – we will immediately remove your name from the mailing list for this study.

2. Call the Ontario Newborn Screening Program [xxx xxx-xxxx ext. xxxx]. Leave a voicemail message with the identification number that appears on the front page of your questionnaire. We will immediately remove your name from the mailing list for this study.

3. Ignore this letter. The newborn screening program will contact you up to 5 times in total about this study. This is done to try to get as many responses as possible so that we can be more confident of the results. If you ignore all five contacts your name will be removed from the mailing list for this study.

Risks and Benefits of Participating in this Study:

There are no specific risks to you or your infant by your participation in this study.

There are no specific benefits to you by participating in this study. However, the knowledge we gain from this study will be useful to the Ontario Newborn Screening Program. We hope to use the results to improve the ways in which we communicate with parents.

Voluntary Participation and Withdrawing from this Study:

You are under no obligation to participate in this study. Your decision to participate or not will not affect any future care your infant may receive from CHEO. You are free to withdraw from the study at any time.

Confidentiality:

Your personal information will be kept strictly confidential. You will not be identified to anyone outside the Ontario Newborn Screening Program. Only grouped results will be included in any publication or presentation of this study.

The researchers we are working with at the University of Ottawa will have access to the questionnaires from this project. If you choose to participate, your name or other identifying information will not appear on your completed questionnaire. The identification number on the front of your questionnaire will be used only by the Ontario Newborn Screening Program to track whether you have responded to the study. This is done so that we know whether to send you a reminder letter. We also need to keep track of the total number of responses.

When your questionnaire is received, your replies will be entered onto a computer database for analysis. The computer file will not contain your name or other identifying information.

The questionnaires will be kept in a secure location. They will be accessible only to the researchers of this study. Once the study is complete, and no further analyses are required, the identification numbers will be permanently removed from computerized data so that they cannot be linked with individual participants.

Questions:

If you have any questions or would like to speak with the researchers of this study, you are free to call Dr. Pranesh Chakraborty at xxx-xxx-xxxx ext. xxxx, or Dr. Beth Potter at xxx-xxx-xxxx ext. xxxx. They will be happy to talk to you without any obligation on your part.

If you would like more information about newborn screening in general, please visit the Ontario Newborn Screening Program's website at:
http://www.newbornscreening.on.ca

This study has been reviewed and approved by the CHEO Research Ethics Board. The Research Ethics Board (REB) is a committee of the hospital that includes individuals from different professional backgrounds. The Board reviews all research that takes place at or through the hospital and its programmes or services. Its goal is to ensure the protection of the rights and welfare of people participating in research. The Board's work is not intended to replace a parent or child's judgement about what decisions and choices are best for them. You may contact the Chair of the Research Ethics Board, for information regarding patient's rights in research studies at xxx-xxx-xxxx ext. xxxx, although this person cannot provide any health-related information about the study.

(Valid until July 6, 2010)

Note: this post-card will be placed in an envelope to be mailed to potential participants, so that the information is kept confidential.
Front of post-card:

[Date]
Dear [name of potential participant] [ID number]
A few days ago you should have received an invitation to participate in a survey concerning mothers' views and experiences with newborn screening.
If you have already completed the questionnaire, I would like to express my sincere thanks. If you have not had the opportunity to complete the survey, but would like to, I encourage you to do so today. Your views are important to this research, which we hope will ultimately contribute to an improved education process.
If you did not receive a survey or your package was misplaced, please contact the Ontario Newborn Screening Program so that I can send you a package today.

Sincerely,
Dr. Pranesh Chakraborty
Laboratory Director/Ontario Newborn Screening Program
[xxx-xxx-xxxx ext. xxxx]

Back of post-card:

Appendix 4: Questionnaire

Mothers' Views and Experiences With Newborn Screening

Study Identification Number

This research project is designed to develop better education about newborn screening. This questionnaire is about where parents get their information about newborn screening. It is not being used to evaluate any screening your child might have had.

This questionnaire asks questions about:

- your pregnancy with your youngest child
- your own experiences and opinions about newborn screening education
- what you understand about newborn screening
- information about you.

We would like you to try to answer all the questions.

All your answers will be kept confidential. Your name is not on the questionnaire.

If you have any questions or concerns about this project, please contact Dr. Pranesh Chakraborty at: xxx-xxx-xxxx ext. xxxx

If you would like more information about newborn screening in general, please visit the Ontario Newborn Screening Program's website at: http://www.newbornscreening.on.ca or call xxx-xxx-xxxx

If you require specific information regarding your infant's health, please contact your family physician or your child's pediatrician.

Please check any of the following statements that apply to you (you may check more than one):

- ❑ I do not have children
- ❑ My youngest child received a positive (abnormal) screening test result
- ❑ My youngest child is adopted
- ❑ My youngest child is older than 6 months
- ❑ I do not wish to participate in this study

If you have checked any of the above then you are unable to participate in this study. In this case, please leave the rest of the questionnaire blank and return it in the envelope provided. Thank you very much for your time.

Section 1: Your Children and Your Pregnancy Experience

1. **How many children do you have?**

   _____

2. **What is the age of your <u>youngest</u> child (or children in the case of twins or triplets)?**

   _____

3. **Where was your <u>youngest</u> child born?**

   - ❑ In a hospital
   - ❑ At home
   - ❑ Other (*please specify*): _____

4. **Which health professional provided the <u>most</u> care during your pregnancy with your <u>youngest</u> child?**

   - ❑ Obstetrician
   - ❑ Family physician
   - ❑ Midwife
   - ❑ Nurse
   - ❑ Other *(please specify)*: _____

5. **Thinking back to your <u>youngest child,</u> did you receive information about pregnancy, childbirth, and/or newborn care from (check one response for each source):**

	Yes	No	Don't know
Prenatal class	❑	❑	❑
Prenatal (before birth) health professional(s)	❑	❑	❑
Post-partum (after birth) health care professional(s)	❑	❑	❑
Family members	❑	❑	❑
Friends	❑	❑	❑
Internet sites (websites)	❑	❑	❑
Television or radio	❑	❑	❑
Books	❑	❑	❑
Other *(please specify):*_____	❑	❑	❑

6. **What would you say was your <u>most</u> important source of information about pregnancy, and/or childbirth?**

   _____

7. **What would you say was your <u>most</u> important source of information about newborn care?**

   _____

Section 2: Your Experiences with Newborn Screening

Newborn screening (also known as the "heel prick test") consists of analyzing blood taken from an infant's heel. Newborns receive this test in the first few days of life to check for certain rare but serious diseases.

8. Before today, had you heard of newborn screening (the "heel prick test")?

- ❑ Yes
- ❑ No
- ❑ Don't know

9. Do you recall your youngest child receiving newborn screening (the "heel prick test")?

- ❑ Yes
- ❑ No
- ❑ Don't know

10. Thinking back to your pregnancy and the newborn period for your youngest child, do you recall receiving any information about newborn screening?

- ❑ Yes
- ❑ No → If 'NO' or 'Don't know' (cannot recall receiving any information about newborn screening), please skip to **question 16**.
- ❑ Don't know →

11. If YES to question 10, thinking back to your pregnancy and the newborn period for your youngest child, did you receive information about newborn screening at any of the following times (check one response for each time)?

	Yes	No	Don't know
Before childbirth (during pregnancy)	❑	❑	❑
After birth, before newborn screening (heel prick) test	❑	❑	❑
At the time of the heel prick test	❑	❑	❑
After the heel prick test	❑	❑	❑

12. If YES to question 10, thinking back to your pregnancy and the newborn period for your youngest child, did you receive information about newborn screening from any of the following sources (check one response for each source)?

	Yes	No	Don't know	Not applicable
Prenatal class	❑	❑	❑	❑
Obstetrician	❑	❑	❑	❑
Family physician	❑	❑	❑	❑
Midwife	❑	❑	❑	❑
Pediatrician	❑	❑	❑	❑
Nurse	❑	❑	❑	❑
Family members	❑	❑	❑	❑
Friends	❑	❑	❑	❑
Internet sites (websites)	❑	❑	❑	❑
Television or radio	❑	❑	❑	❑
Books	❑	❑	❑	❑
Other (*please specify*): _____	❑	❑	❑	❑

13. If YES to question 10, did you receive information about newborn screening in any of the following ways (check one response for each item)?

	Yes	No	Don't know
Personal conversation with a health professional	❑	❑	❑
Group education (e.g., in prenatal class)	❑	❑	❑
Information sheet given at time of heel prick	❑	❑	❑
Other written information (e.g., pamphlet)	❑	❑	❑
Ontario Newborn Screening Program website	❑	❑	❑
Other *(please specify):*_____	❑	❑	❑

14. If YES to question 10, do you remember being told about <u>any</u> of the following aspects of newborn screening (check one response for each item)?

	Yes	No	Don't know
The purpose of the newborn screening test	❑	❑	❑
The importance of having your baby screened	❑	❑	❑
How the screening would be done	❑	❑	❑
Whether you would be told if the result was negative (normal)	❑	❑	❑
Whether you would be told if the result was positive (abnormal)	❑	❑	❑
How you would be told if the result was positive (abnormal)	❑	❑	❑
What a negative (normal) result means	❑	❑	❑
What a positive (abnormal) result means	❑	❑	❑
How the results would be stored	❑	❑	❑
How the blood sample would be handled after testing	❑	❑	❑

15. If YES to question 10, how satisfied are you with the information you were given about newborn screening?

- ❑ Very satisfied
- ❑ Somewhat satisfied
- ❑ Neither satisfied nor dissatisfied
- ❑ Somewhat dissatisfied
- ❑ Very dissatisfied

Section 3: Your Opinions about Newborn Screening Education

16. In your opinion, when should parents receive information about newborn screening (check one response for each time)?

	Yes	No	No opinion
During pregnancy, before childbirth	❑	❑	❑
After birth, before newborn screening (heel prick) test	❑	❑	❑
At the time of the heel prick test	❑	❑	❑
After the heel prick test	❑	❑	❑
Other (*please specify*):_____	❑	❑	❑

17. In your opinion, when is the single ***best* time for parents to receive information about newborn screening (please check one only)?**

- ❑ Before childbirth (during pregnancy)
- ❑ After birth, before newborn screening (heel prick) test
- ❑ At the time of the heel prick test
- ❑ After the heel prick test
- ❑ No opinion

18. In your opinion, who do you expect to be the ***most* knowledgeable about newborn screening (please check one only)?**

- ❑ Obstetrician
- ❑ Family physician
- ❑ Midwife
- ❑ Pediatrician
- ❑ Nurse
- ❑ Other (*please specify*): _____
- ❑ No opinion

19. In your opinion, which health professionals should be responsible for providing newborn screening information to parents (check one response for each health professional)?

	Yes	No	No opinion
Obstetrician	❑	❑	❑
Family Physician	❑	❑	❑
Midwife	❑	❑	❑
Pediatrician	❑	❑	❑
Nurse	❑	❑	❑
Other (*please specify*):_____	❑	❑	❑

20. In your opinion, who should be *most* responsible for providing parents with information about newborn screening (please check one only)?

- ❑ Obstetrician
- ❑ Family physician
- ❑ Midwife
- ❑ Pediatrician
- ❑ Nurse
- ❑ Other (*please specify*): _____
- ❑ No opinion

21. In your opinion, which of the following are important ways for parents to receive information about newborn screening (please check one response for each item)?

	Yes	No	No opinion
Personal conversation with a health professional	❑	❑	❑
Group education (e.g., in prenatal class)	❑	❑	❑
Information sheet given at time of heel prick	❑	❑	❑
Other written information (e.g., pamphlet)	❑	❑	❑
Ontario Newborn Screening Program website	❑	❑	❑
Other *(please specify):*_____	❑	❑	❑

22. In your opinion, which would be the *single best* way for Ontario parents to receive information about newborn screening (please check one only)?

- ❑ Conversation with a health professional
- ❑ Group education (e.g., in prenatal class)
- ❑ Information sheet given at time of heel prick
- ❑ Other written information (e.g., pamphlet or fact sheet)
- ❑ Ontario Newborn Screening Program website
- ❑ Other *(please specify):*_____

23. In your opinion, how important is it for each of the following aspects to be included in newborn screening education for Ontario parents (check one response for each item)?

	Very important	Somewhat Important	Not Important	No Opinion
The purpose of the newborn screening test	❑	❑	❑	❑
The importance of having your baby screened	❑	❑	❑	❑
How the screening will be done	❑	❑	❑	❑
Whether you will be told if the result is negative (normal)	❑	❑	❑	❑
Whether you will be told if the result is positive (abnormal)	❑	❑	❑	❑
How you will be told if the result is positive (abnormal)	❑	❑	❑	❑
What a negative (normal) result means	❑	❑	❑	❑
What a positive (abnormal) result means	❑	❑	❑	❑
How the results will be stored	❑	❑	❑	❑
How the blood sample will be handled after testing	❑	❑	❑	❑

24. Is there anything else that you believe parents should be told about newborn screening in Ontario?

- ❑ Yes
- ❑ No → If 'NO' then please skip to **question 25.**

If 'Yes' please indicate in the space below anything else that you think is important for parents to know:

_____

_____

Section 4: Knowledge about newborn screening

We are interested in finding out what mothers know about newborn screening. These questions will provide useful information for evaluating the current newborn screening education process. The information will also help the Ontario Newborn Screening Program to plan their education initiatives.

25. The <u>main</u> purpose of newborn screening is to identify infants who may have a rare disease so that (please check one only):

- ❑ Parents will know what to expect when their infant begins to show disease symptoms
- ❑ Treatment can begin right away to prevent serious health problems
- ❑ Other family members can be tested to see if they have the disease
- ❑ Don't know

26. To be <u>most</u> accurate, a sample of blood should be taken from the baby:

- ❑ Within one hour after birth
- ❑ At least 24 hours after birth
- ❑ More than one week after birth
- ❑ Don't know

27. A healthcare provider will contact you if your infant's newborn screening test results are positive (abnormal).

- ❑ True
- ❑ False
- ❑ Don't know

28. Your infant may receive a positive (abnormal) newborn screening test result even if he or she does not have a disease.

- ❑ True
- ❑ False
- ❑ Don't know

29. You may choose not to have your infant tested.

- ❑ True
- ❑ False

Section 5: Questions About You

30. How old are you?

- ❑ 19 years or younger
- ❑ 20-24 years
- ❑ 25-29 years
- ❑ 30-34 years
- ❑ 35-39 years
- ❑ 40 years or older

31. What is your current marital status?

- ❑ Single, never married
- ❑ Married or living with partner
- ❑ Divorced/Separated
- ❑ Widowed

32. What is the highest level of education you have completed?

- ❑ None
- ❑ Elementary school
- ❑ Some high school
- ❑ High school diploma
- ❑ Community college, technical college, or CEGEP
- ❑ University degree-undergraduate
- ❑ Professional degree (e.g. education)
- ❑ University degree- graduate or higher
- ❑ Other education or training; please specify:

  _____

33. Are you a healthcare professional?

- ❑ Yes
- ❑ No → If 'NO' (you are not a healthcare professional), or 'Don't know', then please skip to **question 36**.
- ❑ Don't know → If 'NO' (you are not a healthcare professional), or 'Don't know', then please skip to **question 36**.

34. If YES to question 33, which of the following best describes your role?

- ❑ Physician
- ❑ Nurse

- ❑ Midwife
- ❑ Other, please specify (example: physiotherapist):_____

35. If YES to question 33, do you routinely work with pregnant women or new parents in your clinical practice?

- ❑ Yes
- ❑ No

36. What is your language most often spoken at home?

- ❑ English
- ❑ French
- ❑ Chinese
- ❑ Dutch
- ❑ German
- ❑ Greek
- ❑ Italian
- ❑ Japanese
- ❑ Korean
- ❑ Polish
- ❑ Portuguese
- ❑ Punjabi
- ❑ Spanish
- ❑ Other (*please specify*): _____

Thank you for your participation.
Please return the questionnaire in the envelope provided.

Appendix 5: Participant information form, pilot questionnaire and letter of approval from Ottawa Hospital Research Ethics Board

Mothers' Views and Experiences with Newborn Screening

Investigators: Dr. Beth Potter, Makda Araia and Dr. Brenda Wilson from the University of Ottawa; and Dr. Pranesh Chakraborty and Christina Honeywell from the Ontario Newborn Screening Program

Questionnaire Survey

PARTICIPANT INFORMATION FORM

You are invited to participate in a **pilot study**. This participation information form describes the study and outlines what will be asked of you if you decide to take part.

Purpose of this Pilot Study:

We are interested in testing a questionnaire that will be used in a larger study about Ontario mothers' views and experiences with newborn screening. The main goal of this pilot study is to identify potential problems with the questionnaire. The results from this study will be used to make changes to improve the quality of the questionnaire.

Participants:

This pilot study will include approximately 30 pregnant women who are currently attending prenatal classes at the Ottawa Hospital.

Procedures:

This pilot study consists of two (**2**) questionnaires. The main study questionnaire is the one we are evaluating for the larger study. The shorter questionnaire asks your opinion about the main questionnaire. It will take about 30 minutes in total to complete both questionnaires.

If you would like to participate, you may do so by completing both questionnaires and returning them to the study investigators. You may return the questionnaires in person at the end of your prenatal class, or later by mail. A postage-paid envelope will be provided for you in case you choose to mail the questionnaires.

We understand that you may prefer not to take part in this study. If this is the case you may either let us know in person, or simply dispose of the questionnaires.

Risks of Participating in this Study:

There are no specific risks to you by your participation in this study.

Benefits of Participating in this Study:

There are no specific benefits to you by participating in this study. However, the knowledge we gain from this study will be useful to the Ontario newborn screening program in improving the ways in which we communicate with parents.

Voluntary Participation and Withdrawing from this Study:

You are under no obligation to participate in this study. Your decision to participate or not will not affect any future care you or your family receives at the Ottawa Hospital. You are free to withdraw from the study at any time.

Confidentiality:

We will not ask you to provide any identifying information for this pilot study. Participation is anonymous. Completed questionnaires will be securely stored in a locked cabinet by the principal investigator, Dr. Beth Potter. The data collected for this study will be kept for 15 years after termination of the study.

The Ottawa Hospital Research Ethics Board and the Ottawa Health Research Institute may review your relevant study records for audit purposes. If you have any questions about your rights as a research participant, you may contact the Chairman of the Ottawa Hospital Research Ethics Board at xxx-xxx-xxxx, ext. xxxxx.

Questions:

If you have any questions or would like to speak an investigator of this study, you are free to call Dr. Beth Potter at xxx-xxx-xxxx ext.xxxx. She will be happy to talk to you without any obligation on your part.

If you would like more information about newborn screening in general, please visit the Ontario Newborn Screening Program's website at:
http://www.newbornscreening.on.ca

(Valid until May 5, 2010)

Newborn Screening Education: A Survey of Ontario Mothers

Pilot Study: Short Questionnaire

This short questionnaire is part of a pilot study. The pilot study will be used to test a questionnaire about Ontario mothers' views and experiences with newborn screening.

This short questionnaire asks your opinion about the main study questionnaire. Please try out the main questionnaire before you answer this short questionnaire. We are interested in knowing the amount of time it takes to complete the main study questionnaire. We also want to know about questions on the main questionnaire that are difficult to answer so that we can change them.

If you have any questions or concerns about this pilot study, please contact Dr. Beth Potter at: (xxx) xxx-xxxx ext. xxxx

If you would like more information about newborn screening in general, please visit the Ontario Newborn Screening Program's website at: http://www.newbornscreening.on.ca or call [xxx-xxx-xxxx ext. xxxx]

Newborn Screening Education: A Survey of Mothers

We hope that you choose to participate in this study. If you are unable to participate in this study for any reason, please leave the rest of the questionnaire blank and return it in the envelope provided. Thank you very much for your time.

Section 1: Your Opinions about the Newborn Screening Questionnaire

1. Were you able to complete the questionnaire on Newborn Screening?

☐ Yes → If 'YES,' then please skip to **question 3**.
☐ No

2. If NO to question 1, please indicate which of the following best describes why you were *not* able to complete the questionnaire:

☐ I do not wish to participate
☐ I experienced some difficulty answering the questions
☐ I did not have enough time to complete the questionnaire
☐ Other (please specify):

3. If YES to question 1, how long did it take you to complete the questionnaire?

☐ Less than 10 minutes
☐ At least 10 minutes but less than 20 minutes
☐ At least 20 minutes but less than 30 minutes
☐ More than 30 minutes (please specify how long it took): _____minutes
☐ Don't know

Please answer the next few questions even if you were unable to complete the questionnaire.

4. **In your opinion, were ***most*** questions clear, and easy to understand?**

   ☐ Yes
   ☐ No
   ☐ Don't Know

5. **If NO to question 5, which questions would you say were the most difficult to answer?**

_____
_____

For questions 6-10, please indicate how strongly you ***agree*** or ***disagree*** with each of the following statements about the questionnaire. If you **DISAGREE** or **STRONGLY DISAGREE**, then please explain why in space provided.

6. **How strongly do you agree or disagree with the following statement; "The design/layout of the questionnaire is attractive."**

   ☐ Strongly agree
   ☐ Agree
   ☐ Disagree
   ☐ Strongly Disagree

   If 'disagree' or 'strongly disagree', please briefly explain in space below:
   _____
   _____
   _____

7. **How strongly do you agree or disagree with the following statement; "Instructions provided on the questionnaire are clear and easy to follow."**

   ☐ Strongly agree
   ☐ Agree
   ☐ Disagree
   ☐ Strongly Disagree

   If 'disagree' or 'strongly disagree', then please briefly explain in space below:

   _____
   _____
   _____

8. **How strongly do you agree or disagree with the following statement' "The length of time to complete the questionnaire is reasonable."**

   - ☐ Strongly agree
   - ☐ Agree
   - ☐ Disagree
   - ☐ Strongly Disagree

   If 'disagree' or 'strongly disagree', then please briefly explain in space below:
   _____
   _____
   _____

9. **How strongly do you agree or disagree with the following statement; "The ordering of questions make sense and is easy to follow."**

   - ☐ Strongly agree
   - ☐ Agree
   - ☐ Disagree
   - ☐ Strongly Disagree

   If 'disagree' or 'strongly disagree', then please briefly explain in space below:
   _____
   _____
   _____

10. **How strongly do you agree or disagree with the following statement; "Most questions were written in simple language that is easy to understand."**

    - ☐ Strongly agree
    - ☐ Agree
    - ☐ Disagree
    - ☐ Strongly Disagree

    If 'disagree' or 'strongly disagree', then please briefly explain in space below:
    _____
    _____
    _____

The Ottawa Hospital | L'Hôpital d'Ottawa

Research Ethics Board
Conseil d'éthique en recherches
[redacted]
http://www.ohri.ca/ohreb/

Wednesday, May 06, 2009

Dr. Beth Potter
University of Ottawa
Department of Epidemiology and Community Medicine
[redacted]

Dear Dr. Potter:

Re: Protocol # 2009273-01H Newborn Screening Education: A Survey of Ontario Mothers

Protocol approval valid until - Wednesday, May 05, 2010

Thank you for your letter dated April 17, 2009 and for the protocol amendment report dated April 28, 2009. I am pleased to inform you that this protocol underwent expedited review by the Ottawa Hospital Research Ethics Board (OHREB) and is approved. No changes, amendments or addenda may be made to the protocol or the consent form without the OHREB's review and approval.

Approval is for the following documentation:
- English Participant Information Form received April 17, 2009
- English Pilot Study: Short Questionnaire received April 29, 2009
- English Pilot Study: Long Questionnaire received March 26, 2009

The validation date should be indicated on the bottom of all consent forms and information sheets (see copy attached). If the study is to continue beyond the expiry date noted above, a Renewal Form should be submitted to the OHREB approximately six weeks prior to the current expiry date. If the study has been completed by this date, a Termination Report should be submitted.

The Ottawa Hospital Research Ethics Board is constituted in accordance with, and operates in compliance with the requirements of the Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans; Health Canada Good Clinical Practice: Consolidated Guideline; Part C Division 5 of the Food and Drug Regulations of Health Canada; and the provisions of the Ontario Health Information Protection Act 2004 and its applicable Regulations.

Yours sincerely,

[redacted]

Raphael Saginur, M.D.
Chairman
Ottawa Hospital Research Ethics Board

Encl.

/cb

Appendix 6: Outline and summary of results for data cleaning, and univariate results for re-coded variables

Decision rules for inconsistent/incorrect responses:

a) **Exclusion 1**: 'Do not have children'

-If subject(s) selected exclusion criteria 1, then proceeded to respond to questions about her/their youngest child, then will include questionnaire(s) in the study (assumption here is that participant(s) made an error)

b) **Exclusion 2**: 'My youngest child received a positive (abnormal) result'

If subject(s) selected exclusion criteria 2 then will excluded all responses from the study, if it is determined that her/their youngest child actually received a positive result (to check with ONSP). If it is found that the child did not receive positive 'blood spot' screening, then assume that participant misunderstood the question (mother may be confused with other screening/blood tests), and responses will be included in the study.

c) **Exclusion 3**: 'My youngest child is adopted'

If subject selected exclusion 3 then will exclude her/their responses from the study. We do not have sufficient information to confirm this, so will assume that this information is correct.

d) **Exclusion 4**: 'My youngest child is older than 6 months'

If subject selected Exclusion #4 then will cross-check with question #2 and then with ONSP to verify. If it is determined that the child is older than 6 months then his/her mother(s)' responses will be excluded from the study.

e) **Exclusion 5**: 'I do not wish to participate in this study'

- If subject(s) selects criteria#5, but then proceeds to complete the questionnaire then her/their response(s) will be included in this study (will assume that this was an error).

f) **Question 10**- 'do you recall receiving any information about your youngest child' (skip pattern)

- If subject(s) respond 'No' or 'don't know', but answer 'yes' to questions 11-14 (and indicate level of satisfaction for question 15), then will assume that participant answered question 10 incorrectly (i.e.: mother must have received information about NBS to be able to respond to questions 11-15) and responses will be recorded as is. If subject(s) responds 'No' or 'don't know' to question 10 and responded 'No' or 'don't know' to questions 11-14, then her/their responses will be coded 77 (not applicable) (will assume that initial response is correct, and that respondent(s) did not understand the skip pattern).

g) **Question 11**- 'Is there anything else that you believe parents should be told about newborn screening in Ontario?'

If subject(s) responded 'No' but provided their feedback on what information they feel parents should know, then will re-code question 24 as '1' (Yes) (will assume mistakenly selected 'No').

h) **Question 33**- 'Are you a healthcare professional?'

- If subject selected 'No' or 'Don't know' but provided an answer for questions 34 and 35 about their role as a healthcare professional, then will re-code question 33 as '1' (yes) (assume initial response is incorrect, that the individual is in fact a HCP).

Missing data:

1. Determine the proportion of cases with missing data for each variable
2. For key variables compare people with non-missing vs. missing on demographic variables (age and education), to determine likely biases resulting from missing data.
3. If level of missing data <5% then case-wise deletion
4. What to do if level of missing data is >10%?

Out-of-range responses:

1. Cross check data entry (check for discrepancies between coders, refer to table 1 and 2 for list of codes for each question).

Data Entry Codes:

Table 1

Yes	**1**
No	**2**
Don't know	**88**
Other	**66**
Not applicable	**77**
No opinion	**55**
Missing	**99**

Table 2

Variable name	Description	Codes
Exclusion 1-5	Exclusion criteria; if code=1 for any, then Q1-Q36 = 77	1, 2 or 77
Q1	Number of children	1-5 or 99 (if blank)
Q2	Age of youngest child	0-6 (in months) or 99
Q3	Birth place	1, 2, 66, 99
Q4 & Q4other	Health care providers	1 , 2, 3, 4, 66, 99
Q5a-Q5i other	Info about childbirth etc…	1, 2, 88, 99
Q6a-Q6i other	Important source of info about pregnancy/childbirth	1, 2, 99
Q7a-Q7i other	Important source of info about newborn care	1, 2, 99
Q8	Do you recall hearing about NBS?	1, 2, 88, 99
Q9	Do you recall your child receiving NBS?	1, 2, 88, 99
Q10	Receiving info on NBS [if =2 or 88, then Q12-15= 77]	1, 2, 88, 77, 99
Q11a-Q11d	When was info about NBS received?	1, 2, 77, 88, 99
Q12a-Q12l other	Who provided NBS information?	1, 2, 77, 88, 99
Q13a- Q13f other	How was NBS info provided?	1, 2, 77, 88, 99
Q14a- Q14j	What information was provided?	1, 2, 77, 88, 99
Q15	Level of satisfaction with NBS information	1, 2, 3, 4, 5, 77, 99
Q16a-Q16e other	When should NBS info be given?	1, 2, 55, 99
Q17	When is the ***best*** time to receive NBS information?	1, 2, 3, 4, 55, 99
Q18 -Q18other	Who is most knowledgeable about NBS?	1, 2, 3, 4, 5, 55, 66, 99
Q19a- Q19f other	Which HCP should provide NBS info?	1, 2, 55, 99
Q20-Q 20a other	Which HCP is most responsible for providing NBS info?	1, 2, 3, 4, 5, 55, 66, 99
Q21a- Q21f other	What are important ways to receive NBS info?	1, 2, 55, 99
Q22- Q22a other	What is the single best way to receive NBS info?	1, 2, 3, 4, 5, 66, 99
Q23a – Q23j	Level of importance of NBS information	1, 2, 3, 55, 99
Q24	Any other info that should be provided to parents	1, 2, 99
Q24a	Open-ended	text
Q25	Knowledge Question = purpose of NBS	1, 2, 3, 88, 99
Q26	Knowledge Question= When should screening occur?	1, 2, 3, 88, 99

Disposition status

Number in original sampling frame	1712
Responded by mail	714
Responded by web	36
Returned to sender (not at address in NBS database)	128
Active refusal	66
Found to be ineligible	2
Unknown outcome	766
Number of respondents who completed more than 80% of applicable & crucial questions (complete response)*	748
Number of respondents who completed 50-80% of applicable and crucial questions * (partial response)	1
Number of respondents who completed less than 50% of applicable and crucial questions * (break-off)	1

* Crucial questions are all questions excluding those in section 5 (demographics).

Problems identified through data cleaning

Incorrect selection of exclusion criteria

Several participants (n=50) selected one of the 5 exclusion criteria but provided responses to subsequent questions (as opposed to leaving the questionnaire blank as instructed). Three mothers indicated that they did not have children at the time of the study (exclusion criterion 1) but later responded to questions about their youngest child (for example indicated the age of their youngest child). It was therefore determined that mothers responded incorrectly to the exclusion question and so their responses were included in the analyses. Forty-one mothers indicated that their youngest child received a positive (abnormal) screening test result (exclusion criterion 2). All 41 mothers provided responses to subsequent questions in the questionnaire. This prompted NSO to check the original sampling frame and ensure that the sample was selected from mothers with negative results. After affirming that the sample was selected from a group of mothers whose infants were known to have a negative screening results, it was assumed that an error was made (incorrectly selecting exclusion criterion 2) and so responses from these 40 mothers were included for analysis (one mother

was excluded because she had also indicated that her child was older than 6 months). Eight mothers indicated that their youngest child was older than 6 months (exclusion criterion 4). After cross-checking these responses with responses from question 2 (age of youngest child), it was determined that 7 mothers incorrectly selected exclusion 4 (they indicated that their child was actually less than 6 months of age in question 2) and so their responses were included in subsequent analyses.

Errors in following skip patterns

Investigators also noted errors with responding to questions that involved skip patterns (for example questions 10-15 and 33-35). In question 10, mothers were asked to indicate whether they recalled receiving information about NBS during pregnancy or the newborn period with their youngest child. Mothers were instructed to skip questions 11 to 15 if they selected 'no' or 'don't know' to question 10. Four mothers indicated that they did not receive or did not know whether they received information about NBS but responded to latter questions about the information they received by consistently responding either 'no' or 'don't know' to any of questions 11-15. Here, the original responses to question 10 were kept, but responses to questions 11-15 were set to missing. There was one mother who answered 'no' or 'don't know' to question 10 but provided an affirmative response to at least one part of questions 11-15. In this case, we assumed an error was made in question 10 and changed the response to 'yes'. Other errors related to skip patterns involving questions 33-35 were identified. Here, each participant was asked to identify whether she was a healthcare professional. Those individuals who answered 'no' or 'don't know' were instructed to leave questions 34 and 35 blank. A few mothers (n=6) responded 'no' or 'don't know' to question 33, left question 34 blank but responded 'no' to question 35. Again, we assumed that mothers erroneously responded to question 35 (a probable reason for this error being that this question was located

on the next page of the questionnaire, and so may have been interpreted as not being related). In these cases the responses to question 35 were set to missing.

Unclear responses

A coding scheme was used to categorize all responses to open-ended questions. There were several instances (n=77) where mothers provided answers (for example incomplete sentences or responses) which could not be easily categorized/coded. This was particularly true for question 6 where mothers were asked to describe their most important source of information about pregnancy, and/childbirth (n=18). These responses were coded as missing.

Multiple responses

Multiple responses to questions where a single answer was requested were found to be one of the most common types of errors (occurred 525 times), particularly among open-ended questions or questions that allowed mothers the opportunity to provide additional or 'other' responses (ex.: question 5). These types of errors are of particular concern (impeding comparability among respondents) considering that online respondents did not have an opportunity to provide multiple responses to questions where a single response was requested (ex.: question 17). In an attempt to prevent substantial loss of data multiple responses were included in the analyses for questions 6 and 7, where a large number of multiple responses were given (question 6, n=155; question 7, n=178). Another exception was question 36 (language most often spoken at home) where responses of English and French were not excluded (n=19). For all other questions multiple responses were set to missing.

Missing data

Missing data was the most common type of error identified in this study (occurring at least once for each variable). The general approach to item-missing data was to exclude

participants with missing data on any variable that contributed to a particular analysis. This strategy did not result in excessive exclusions for most analyses since most variables had fewer than 5% item missing data. We identified a number of questions (n=6) which were more likely to have numerous missing responses on all or part of the question. These questions generally included multiple categories (which are mutually exclusive) whereby mothers were required to select a response (ex.: 'yes', 'no', or 'don't know') for each item for that particular question. For example in question #5, mothers were asked to indicate whether they recalled receiving information about pregnancy, childbirth or newborn care from a variety of sources (e.g., prenatal class, prenatal/post-natal healthcare providers…; see appendix 4). Upon further examination it appeared as though mothers may have misinterpreted the instructions for these questions (they were asked to check one response for each item), and treated these questions as 'check-all-that-apply'; whereby a response is provided only if it applicable, leaving non-applicable items blank. To test this assumption, we decided to create new variables whenever it was determined that a variable had more than 10% of responses missing. These new variables (n=6) were recoded such that item missing responses were set to 'no' if the participant responded 'yes' to any of the other categories for that question (responses of 'no' or 'don't know' were unchanged). Descriptive statistics were conducted on both old and new variables, and results were compared. In all cases treating the variables as 'check-all-that-apply' resulted in fewer item missing responses for each variable, but also resulted in slight increases and decreases (≤5%) in the proportion responding 'yes' and 'no' respectively (the proportion of those who responded 'don't know' remained unchanged). For example in question 11 where mothers were asked to indicate when they recalled receiving information about NBS, there were 54 missing responses to the first

response category (before childbirth-during pregnancy). Upon recoding there were only 11 missing responses for that item. As a result of the substantial decrease in the proportion of missing data following this approach, these new variables were used in all subsequent analyses (see results for re-coded variables).

Results for re-coded variables

	Frequency before re-coding			Frequency after re-coding		
Question	**Yes**	**No**	**Missing**	**Yes**	**No**	**Missing**
	%	**%**	**n**	**%**	**%**	**n**
11A	35.5	29.5	54	33.4	33.6	11
11 B	45.4	18.8	54	42.9	23.3	13
11 C	52.3	12.2	60	49.2	17.4	16
11 D	30.9	31.0	83	28.3	36.9	21

Appendix 7: Letter of Support from Newborn Screening Ontario

CHEO Children's Hospital of Eastern Ontario
Centre hospitalier pour enfants de l'Est de l'Ontario

Ontario Newborn Screening Program
Programme de dépistage des nouveaux-nés de l'Ontario

October 17, 2008

Dr. Beth Potter
Epidemiology & Community Medicine
[redacted]

Dear Dr. Potter,

I am writing you this letter to confirm our support for your graduate student's (Makda Araia) research project entitled "Newborn Screening Education: A Survey of Ontario Mothers." As we discussed, this is an area of interest for the Ontario Newborn Screening Program and this research is both timely and will generate novel knowledge that will inform further decisions at the Program. We will send the surveys for this study and track responses, as well as participate in the overall conduct and analysis for this research project.

Sincerely,

[redacted]

Dr. Pranesh Chakraborty, MD, FRCPC, FCCMG
Metabolic Geneticist and Laboratory Director
Department of Genetics / Ontario Newborn Screening Program
Children's Hospital of Eastern Ontario
[redacted]
www.newbornscreening.on.ca

[redacted] www.cheo.on.ca
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Faire la différence dans la vie des enfants, des adolescents et des familles

Appendix 8: Letter of approval from Children's Hospital of Eastern Ontario Research Ethics Board

CHEO Children's Hospital of Eastern Ontario
Centre hospitalier pour enfants de l'est de l'Ontario

CHEO RESEARCH ETHICS BOARD APPROVAL – EXPEDITED REVIEW

Principal Investigator:	Dr. Pranesh Chakraborty
Proposal Number:	#09/31X
Protocol Title:	Newborn Screening Education: A survey of Ontario Mothers
Department or PSU:	Ontario Newborn Screening Program, Genetics PSU
Approval date:	July 6, 2009
Valid Until:	July 6, 2010
Documents reviewed and approved:	Research protocol submitted March 17, 2009

This is to notify you that the Children's Hospital of Eastern Ontario Research Ethics Board has granted approval to the above named research study on the date noted above. Your project was reviewed under the expedited stream, which is reserved for projects that involve no more than minimal risk to human subjects.

During the course of the research, no deviations from, or changes to, the protocol or consent form may be initiated without prior written approval from the REB. Further, investigators are asked to report the following to the REB:

- Proposed changes to the study procedures (including the recruitment strategy, inclusion criteria, etc.);
- Concerns or issues that arise in conducting the research;
- Changes to the consent documents and advertisement notices;
- Changes to the investigators who assume responsibility for the study; &
- An annual report.

Wishing you success in your project.

Regards,

[redacted]

Dr. Carole Gentile, C.Psych.
Chair, Research Ethics Board

CG/smeh 03/07/2009
c.c. CHEO RI Administration
Beth Potter

This is an official document. Please retain the original for your file *version 11/2003*

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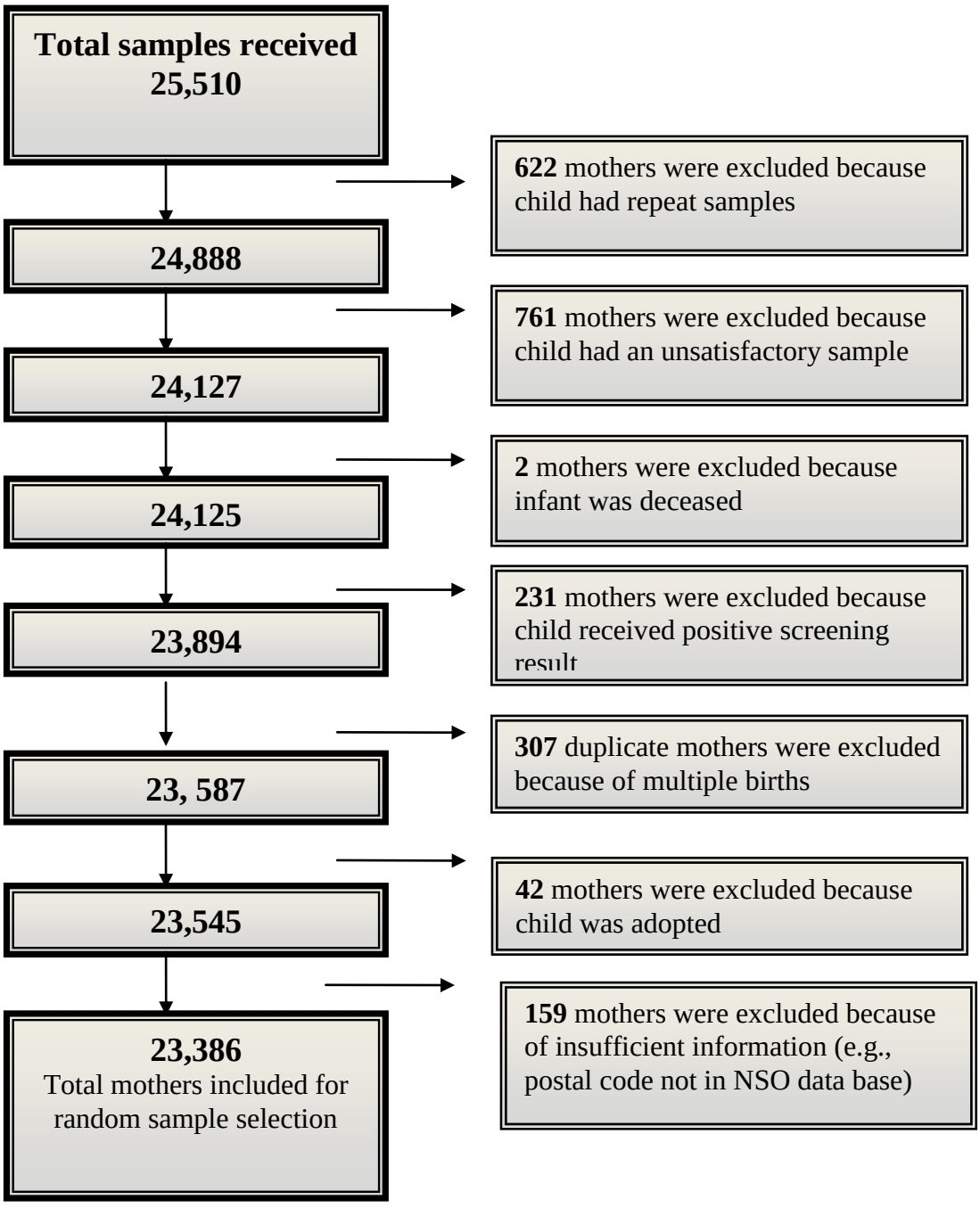
Appendix 9: Figure 1, Breakdown of sample selection



Appendix 10: Univariate results for open-ended questions

Question 6- Most Important source of Information about Pregnancy, and/or Childbirth (N=721)

1. Health care professional(s) (n=239)
2. Books (n=130)
3. Internet (n=79)
4. Family (n=24)
5. Experience (n=17)
6. Friends (n=13)
7. Non print media (n<5)
8. Print media-other than books (n<5)
9. Health unit/facility (n<5)
10. Multiple responses (n=155)
11. Don't know (n<5)

Question 7- Most Important source of Information about Newborn care (N=710)

1. Health care professional(s) (n=214)
2. Books (n=86)
3. Family (n=85)
4. Internet (n=44)
5. Prenatal class (n=41)
6. Experience (n=25)
7. Friends/colleagues (n=17)
8. Healthcare unit/facility (n=14)
9. Non-print media (n<5))
10. Print media-other than books (n<5)
11. Post-partum class (n<5)
12. Multiple responses (n=178)
13. Don't know (n<5)

Question 12L (n=26)-Other Sources of NBS information

1) Print media- other than books (n=15)
2) Healthcare professional-other (n<5)
3) School (n<5)
4) Healthcare facility/centre (n<5)
5) Experience (n<5)
6) Non-healthcare caregiver (n<5)

Question 13F (N=4)-Other ways in which Mothers recall receiving NBS information [a]

1) Internet (n<5)
2) Experience (n<5)

3) Friends (n<5)
4) Family (n<5)

[a] Respondent(s) provided more than one response to this question

Question 18A (N=8)-Other HCP who Mothers expect to be most knowledgeable about NBS.

1) Healthcare professional(s)-other (n=5)
2) Nurse practioner (n<5)
3) Prenatal health professional (n<5)
4) Specialist (n<5)

Question 21F (N=3)-Other important ways to receive information about NBS

1) Non-print media (n<5)

Question 24- Other information Mothers believe Parents should be told about NBS (N=127) [a]

[a] Respondents provided more than one response to this question

n=>10

1) Conditions included for screening
2) Next steps after positive result
3) Results should be given whether positive/negative
4) Information about consent/refusal process

n=>5

5) How screening is done
6) Information about false positive/negatives
7) Benefits of screening
8) How/when results are communicated
9) Risk of pain/discomfort/infection
10) Implications of an abnormal result to the child
11) Probability of receiving positive result

n<5

12) Information about any additional resources
13) Definitions of normal/abnormal results
14) Other health information not related to NBS test
15) Policy information on privacy
16) Medication provided during testing
17) Purpose of screening
18) History of NBS
19) Prevention of abnormal results
20) Possibility of follow-up testing
21) Probability of false positives/negatives
22) Policy information on sample storage and use
23) Information on Supplemental testing
24) Costs of screening

25) Conditions not included for testing
26) Next steps after screening
27) How positive results will be communicated
28) Information about other programs
29) Testing is done on all infants

Question 34- Types of healthcare professions (N=97) [a]

1) Physician (n<5)
2) Nurse (n=27)
3) Other (n=67)

[a] 631 respondents were excluded because they had indicated in a previous question (Question 33) that they were not a healthcare professional.

Question 34A (N=67)-Other types of healthcare professions [a]

n>5

1) Personal support worker
2) Technician lab/pharmacy
3) Dental hygienist/assistant
4) Occupational/Physiotherapist
5) Pharmacists
6) Radiation/message therapist
7) Dietician

n<5

1) Administrative clerk
2) Speech language pathologist/audiologist
3) Vetinarian
4) Optician
5) Dentist
6) Psychologist
7) Epidemiologist

[a] 631 respondents were excluded because they had indicated in a previous question (Question 33) that they were not a healthcare professional.

Question 35- Mothers who report routinely working with pregnant women (N=98) [a]

Yes- (n=33) *p*= 0.34 (0.24, 0.44)
No- (n=65) *p*= 0.66 (0.56, 0.76)

[a] 631 respondents were excluded because they had indicated in a previous question that they were not a healthcare professional.

Appendix 11: Bivariate results- Sources of NBS education

Table 25: Bivariate analysis: Proportion of mothers who recalled receiving information about newborn screening: prenatal class by age, education, language most often spoken at home and parity (Question 12)

	Total	Reported receiving NBS info from pre-natal class	
Prenatal class	**n**	**n (%)**	**OR (95%CI)[†]**
Age (years) [a]			
≤24	50	12 (2.4)	Ref
25-29	112	30 (5.9)	1.16 (0.54-2.51)
30-34	208	40 (7.9)	0.75 (0.36-1.57)
≥35	135	16 (3.2)	0.43 (0.19-0.98)[b]
Education [a]			
High school or less	76	13 (2.6)	Ref
College	140	27 (5.4)	1.16 (0.56-2.40)
University or higher	281	57 (20.3)	1.23 (0.64-2.40)
Language most often spoken at home [a]			
English	408	80 (16.6)	Ref
Other	75	13 (2.7)	0.86 (0.45-1.64)
Parity [a]			
Primiparous	240	85 (16.8)	Ref
Multiparous	267	13 (2.6)	0.09 (0.05-0.17)**

* p<0.05, ** p<0.01,[b] p=0.045, [†]unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

Table 26: Bivariate analysis: Proportion of mothers who recalled receiving information about newborn screening: family by age, education, language most often spoken at home and parity (Question 12)

	Total	Reported receiving NBS info from family	
Family	**n**	**n (%)**	**OR (95%CI)** [†]
Age (years) [a]			
≤24	50	8 (16)	Ref
25-29	112	13 (12)	1.27 (0.65-2.49)
30-34	210	17 (8)	0.71 (0.38-1.31)
≥35	136	12 (9)	0.85 (0.43-1.68)
Education [a]			
High school or less	77	12 (16)	Ref
College	143	10 (7)	0.41 (0.17-0.99)[b]
University or higher	280	26 (9)	0.55 (0.27-1.16)
Language most often spoken at home [a]			
English	410	44 (11)	Ref
Other	75	5 (7)	0.59 (0.23-1.55)
Parity [a]			
Primiparous	241	32 (13)	Ref
Multiparous	269	18 (7)	0.47 (0.26-0.86)*

*p<0.05, **p<0.01, [b] p=0.0479, [†]Unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

Table 27: Bivariate analysis: Proportion of mothers who recalled receiving information about newborn screening: friends by age, education, language most often spoken at home and parity (Question 12)

	Total	Reported receiving NBS info from friends	
Friends	**n**	**n (%)**	**OR (95%CI)** [†]
Age (years) [a]			
≤24	49	8 (16)	Ref
25-29	112	16 (14)	1.66 (0.88-3.11)
30-34	209	19 (9)	0.80 (0.44-1.45)
≥35	136	9 (7)	0.54 (0.26-1.14)
Education [a]			
High school or less	76	8 (11)	Ref
College	142	13 (9)	0.86 (0.34-2.17)
University or higher	280	30 (11)	1.02 (0.45-2.33)
Language most often spoken at home [a]			
English	409	44 (11)	Ref
Other	74	5 (7)	0.60 (0.23-1.57)
Parity [a]			
Primiparous	240	34 (14)	Ref
Multiparous	268	18 (7)	0.42 (0.24-0.80)*

*p<0.05, **p<0.01, [†]Unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

Table 28: Bivariate analysis: Proportion of mothers who recalled receiving information about newborn screening: books by age, education, language most often spoken at home and parity (Question 12)

	Total	Reported receiving NBS info from Books	
Books	**n**	**n (%)**	**OR (95%CI)** [†]
Age (years) [a]			
≤24	46	15 (33)	Ref
25-29	107	39 (36)	1.30 (0.83-2.05)
30-34	197	66 (34)	1.14 (0.77-1.68)
≥35	127	32 (25)	0.65 (0.41-1.02)
Education [a]			
High school or less	72	20 (28)	Ref
College	135	41 (30)	1.13 (0.60-2.14)
University or higher	265	88 (33)	1.29 (0.73-2.30)
Language most often spoken at home [a]			
English	392	124 (32)	Ref
Other	69	26 (38)	1.31 (0.77-2.22)
Parity [a]			
Primiparous	228	85 (37)	Ref
Multiparous	251	68 (27)	0.63 (0.43-0.92)*

*p<0.05, **p<0.01, [†]Unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; TV= television; OR= odds ratio CI= confidence interval

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

Appendix 12: Bivariate results-Methods of NBS education

Table 29: Bivariate analysis: Proportion of mothers who reported the following as important ways to receive NBS information: conversation with HCP, by age, education, language most often spoken at home and parity. (Question 21)

	Total	Reported conversation with HCP	
Conversation with a HCP	**n**	**n (%)**	**OR (95%CI)†**
Age (years)			
≤24	68	67 (99)	Ref
25-29	176	163 (93)	0.65 (0.33-1.28)
30-34	296	287 (97)	2.45 (1.15-5.22)*
≥35	204	186 (91)	0.46 (0.24-0.87)*
Education			
High school or less	111	104 (94)	Ref
College	212	197 (93)	0.88 (0.35-2.24)
University or higher	409	391 (96)	1.46 (0.60-3.59)
Language most often spoken at home			
English	566	546 (96)	Ref
Other	140	121 (86)	0.23 (0.12-0.45)**
Parity			
Primiparous	350	334 (95)	Ref
Multiparous	395	370 (94)	0.71 (0.37-1.35)

*p<0.05, **p<0.01, †Unadjusted OR

Abbreviations: HCP= Healthcare professional; NBS= Newborn screening; Info= information; TV= television; OR= odds ratio; CI= confidence interval

Table 30: Bivariate analysis: Proportion of mothers who reported the following as important ways to receive NBS information: group education (i.e.: pre-natal class), by age, education, language most often spoken at home and parity. (Question 21)

	Total	Reported Group education	
Group education	**n**	**n (%)**	**OR (95%CI)†**
Age (years)			
≤24	68	50 (74)	Ref
25-29	176	139 (79)	1.24 (0.82-1.87)
30-34	296	231 (78)	1.20 (0.85-1.70)
≥35	204	146 (72)	0.72 (0.50-1.04)
Education			
High school or less	111	78 (70)	Ref
College	212	162 (76)	1.37 (0.82-2.30)
Undergraduate	409	318 (78)	1.50 (0.93-2.36)
Language most often spoken at home			
English	566	439 (78)	Ref
Other	140	102 (73)	0.78 (0.51-1.18)
Parity			
Primiparous	350	282 (81)	Ref
Multiparous	395	285 (75)	0.63 (0.44-0.88)*

*p<0.05, **p<0.01, †Unadjusted OR

Abbreviations: HCP= Healthcare professional; NBS= Newborn screening; Info= information; TV= television OR= odds ratio; CI= confidence interval

Table 31: Bivariate analysis: Proportion of mothers who reported the following as important ways to receive NBS information: information sheet, by age, education, language most often spoken at home and parity. (Question 21)

	Total	Reported Information sheet	
Information sheet	**n**	**n (%)**	**OR (95%CI)** †
Age (years)			
≤24	67	53 (79)	Ref
25-29	176	143 (81)	0.80 (0.51-1.24)
30-34	296	256 (86)	1.42 (0.94-2.14)
≥35	204	170 (83)	0.96 (0.62-1.49)
Education			
High school or less	111	91 (82)	Ref
College	211	183 (87)	1.44 (0.77-2.69)
University or higher	409	340 (83)	1.08 (0.63-1.88)
Language most often spoken at home			
English	566	488 (86)	Ref
Other	139	104 (75)	0.48 (0.30-0.75)**
Parity			
Primiparous	349	294 (84)	Ref
Multiparous	395	330 (84)	0.95 (0.64-1.41)

*p<0.05, **p<0.01, †Unadjusted OR

Abbreviations: HCP= Healthcare professional; NBS= Newborn screening; Info= information; TV= television OR= odds ratio; CI= confidence interval

Appendix 13: Bivariate results-Knowledge about NBS

Table 32: Bivariate analysis: Proportion of mothers who responded correctly to a knowledge questions about NBS: purpose of screening, by demographics and experience with education (Question 25)

	Total	**Correct response to purpose of NBS**	
	n	**n (%)**	**OR (95%CI)**[†]
Age (years)			
≤24	64	54 (84)	Ref
25-29	169	137 (81)	0.79 (0.37-1.72)
30-34	287	242 (84)	1.00 (0.47-2.10)
≥35	198	156 (79)	0.69 (0.32-1.47)
Education			
High school or less	106	77 (73)	Ref
College	204	168 (82)	1.76 (1.01-3.07)*
University or higher	397	334 (84)	2.00 (1.21-3.31)*
Language most often spoken at home			Ref
English	550	453 (82)	0.80 (0.50-1.29)
Other	133	105 (79)	
Parity			Ref
Primiparous	339	277 (82)	1.06 (0.72-1.55)
Multiparous	379	312 (82)	2.05 (1.39-3.04)**[b]
Received NBS info	583	419 (72)	
Received info pre-natally [a, d]	417	214 (51)	1.98 (1.16-3.36)* [b]
Received info from a HCP [a, **d**]	418	365 (87)	0.94 (0.52-1.69) [b]
Received info from a pre-natal class [a, d]	417	81 (19)	1.37 (0.67-2.81) [b]
Received info from tear-off sheet [a, c, d]	417	287 (69)	0.86 (0.49-1.51) [b]
Recalled receiving information about: [**a, d**]			
Purpose/benefits of screening	438	384 (88)	3.23 (1.65-6.34)** [b]
How screening would be done	413	358 (87)	1.57 (0.82-3.01) [b]
Whether/how results are communicated	293	256 (87)	1.38 (0.83-2.31) [b]
What negative/positive results mean	198	172 (87)	1.15 (0.68-1.95) [b]
Policy info on storage and use	58	50 (86)	1.03 (0.47-2.29) [b]

*p<0.05, **p<0.01, [†]Unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval; HCP= Healthcare professional

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

[b] Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about information received.

[c] The tear-off sheet is a double-sided page of information for parents that is attached to the blood collection card.

[d] All of these questions were separate so responses are not mutually exclusive (e.g.: mothers could report receiving information from both pre-natal class and the tear-off sheet).

Table 33: Bivariate analysis: Proportion of mothers who responded correctly to a knowledge question about when NBS should occur by demographics and experience with NBS education. (Questions 26)

	Total	Correct response to timing of NBS	
	n	n (%)	OR (95%CI)[†]
Age (years)			
≤24	69	49 (71)	Ref
25-29	176	123 (70)	0.95 (0.51-1.75)
30-34	297	227 (76)	1.32 (0.74-2.38)
≥35	205	148 (72)	1.06 (0.58-1.94)
Education			
High school or less	114	76 (67)	Ref
College	213	164 (77)	1.67 (1.01-2.77)*
University or higher	408	300 (76)	1.39 (0.89-2.17)
Language most often spoken at home			
English	567	426 (75)	Ref
Other	141	98 (70)	0.75 (0.50-1.13)
Parity			
Primiparous	351	245 (70)	Ref
Multiparous	395	302 (76)	1.41 (1.01-1.95)*
Received NBS info	546	402 (74)	2.22 (1.58-3.13)** [b]
Received info pre-natally [a, d]	400	185 (46)	0.61 (0.40-0.94)* [b]
Received info from a HCP [a, d]	400	351 (88)	1.67 (1.05-2.66)* [b]
Received info from a pre-natal class [a, d]	399	71 (18)	0.63 (0.38-1.04) [b]
Received info from tear-off sheet [a, c, d]	400	292 (73)	2.65 (1.71-4.13)** [b]
Recalled receiving information about: [a, d]			
Purpose/benefits of screening	457	366 (80)	2.01 (1.06-3.82)* [b]
How screening would be done	429	349 (81)	2.32 (1.36-3.95)* [b]
Whether/how results are communicated	304	246 (81)	1.38 (0.90-2.12) [b]
What negative/positive results mean	207	170 (82)	1.45 (0.93-2.26) [b]
Policy info on storage and use	61	46 (75)	0.80 (0.43-1.51) [b]

*p<0.05, **p<0.01, [†]Unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval; HCP= Healthcare professional

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

[b] Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about information received.

[c] The tear-off sheet is a double-sided page of information for parents that is attached to the blood collection card.

d All of these questions were separate so responses are not mutually exclusive (e.g.: mothers could report receiving information from both pre-natal class and the tear-off sheet).

Table 34: Bivariate analysis: Proportion of mothers who responded correctly to a knowledge question about how NBS results are communicated, by demographics and experience with NBS education. (Questions 27)

	Total	Correct response to communication of results	
	n	n (%)	OR (95%CI)[†]
Age (years)			
≤24	68	51 (75)	Ref
25-29	176	133 (76)	1.03 (0.54-1.97)
30-34	296	219 (74)	0.95 (0.52-1.74)
≥35	203	160 (79)	1.24 (0.65-2.36)
Education			
High school or less	113	82 (73)	Ref
College	212	150 (71)	0.92 (0.55-1.52)
University or higher	406	324 (80)	1.49 (0.93-2.41)
Language most often spoken at home			
English	564	422 (75)	Ref
Other	141	111 (79)	1.25 (0.80-1.95)
Parity			
Primiparous	348	251 (72)	Ref
Multiparous	311	311 (79)	1.43 (1.02-2.00)*
Received NBS info	559	400 (72)	1.69 (1.19-2.40)* [b]
Received info pre-natally [a, d]	398	195 (49)	1.00 (0.65-1.54) [b]
Received info from a HCP [a, d]	398	340 (85)	0.63 (0.37-1.08) [b]
Received info from a pre-natal class [a, d]	397	75 (19)	0.86 (0.50-1.46) [b]
Received info from info sheet [a, c, d]	396	281 (71)	1.63 (1.04-2.55)* [b]
Recalled receiving information about: [a, d]			
Purpose/benefits of screening	454	363 (80)	1.81 (0.95-3.48) [b]
How screening would be done	428	348 (81)	2.27 (1.32-3.89)** [b]
Whether/how results are communicated	303	268 (88)	4.19 (2.65-6.61)** [b]
What negative/positive results mean	205	185 (90)	3.56 (2.12-5.97)** [b]
Policy info on storage and use	61	53 (87)	1.90 (0.87-4.14) [b]

*p<0.05, **p<0.01, [†]Unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval; HCP= Healthcare professional

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

[b] Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about information received.

[c] The tear-off sheet is a double-sided page of information for parents that is attached to the blood collection card.

[d] All of these questions were separate so responses are not mutually exclusive (e.g.: mothers could report receiving information from both pre-natal class and the tear-off sheet).

Table 35: Bivariate analysis: Proportion of mothers who responded correctly to a knowledge question about the possibility of receiving a false positive result, by demographics and experience with NBS education. (Questions 28)

	Total	Correct response to possibility of FP results	
	n	n (%)	OR (95%CI)[†]
Age (years)			
≤24	69	20 (29)	Ref
25-29	176	58 (33)	1.20 (0.66-2.21)
30-34	297	110 (37)	1.44 (0.81-2.55)
≥35	203	74 (36)	1.41 (0.78-2.54)
Education			
High school or less	114	32 (28)	Ref
College	213	61 (29)	1.03 (0.62-1.70)
University or higher	406	167 (41)	1.79 (1.14-2.82)*
Language most often spoken at home			Ref
English	566	197 (35)	1.21 (0.83-1.77)
Other	140	55 (39)	
Parity			
Primiparous	351	127 (36)	Ref
Multiparous	393	135 (34)	0.93 (0.68-1.25)
Received NBS info	260	198 (76)	1.73 (1.23-2.43)** [b]
Received info pre-natally [a, d]	197	100 (51)	1.14 (0.80-1.63) [b]
Received info from a HCP [a, d]	198	177 (89)	0.94 (0.63-1.41) [b]
Received info from a pre-natal class [a, d]	197	45 (23)	1.44 (0.92-2.25) [b]
Received info from tear-off sheet [a, c, d]	196	148 (76)	1.75 (1.17-2.61)** [b]
Recalled receiving information about: [a, d]			
Purpose/benefits of screening	456	184 (40)	1.64 (0.86-3.15) [b]
How screening would be done	428	176 (41)	1.68 (0.99-2.87) [b]
Whether/how results are communicated	302	131 (43)	1.56 (1.07-2.26)* [b]
What negative/positive results mean	206	98 (48)	1.80 (1.25-2.59)** [b]
Policy info on storage and use	61	31 (51)	1.72 (1.00-2.95) [b]

*p<0.05, **p<0.01, [†]Unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval; HCP= Healthcare professional

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

[b] Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about information received.

[c] The tear-off sheet is a double-sided page of information for parents that is attached to the blood collection card.

[d] All of these questions were separate so responses are not mutually exclusive (e.g.: mothers could report receiving information from both pre-natal class and the tear-off sheet).

Table 36: Bivariate analysis: Proportion of mothers who responded correctly to a knowledge question about the right to refuse NBS, by demographics and experience with NBS education (Questions 29)

	Total	Correct response to the right to refuse testing	
	n	n (%)	OR (95%CI)[†]
Age (years)			
≤24	68	20 (29)	Ref
25-29	176	59 (34)	1.21 (0.66-2.22)
30-34	297	102 (37)	1.26 (0.71-2.23)
≥35	205	76 (37)	1.41 (0.78-2.56)
Education			
High school or less	113	27 (24)	Ref
College	213	76 (36)	1.77 (1.06-2.96)*
University or higher	408	152 (37)	1.89 (1.17-3.05)*
Language most often spoken at home			
English	567	207 (37)	Ref
Other	140	40 (29)	0.70 (0.46-1.04)
Parity			
Primiparous	350	113 (32)	Ref
Multiparous	395	144 (36)	1.19 (0.88-1.62)
Received NBS info	256	207 (81)	2.51 (1.75-3.60)** [b]
Received info pre-natally [a, d]	206	115 (56)	1.61 (1.13-2.30)** [b]
Received info from a HCP [a, d]	206	186 (90)	0.61 (0.41-0.91)* [b]
Received info from a pre-natal class [a, d]	197	45 (23)	0.73 (0.46-1.16) [b]
Received info from tear-off sheet [a, c, d]	204	137 (67)	0.91 (0.63-1.34) [b]
Recalled receiving information about: [a, d]			
Purpose/benefits of screening	457	194 (42)	2.21 (1.12-4.36)* [b]
How screening would be done	429	187 (44)	2.28 (1.31-3.96)* [b]
Whether/how results are communicated	304	143 (47)	1.89 (1.30-2.74)** [b]
What negative/positive results mean	207	115 (56)	2.85 (1.97-4.13)** [b]
Policy info on storage and use	61	34 (56)	2.00 (1.17-3.44)* [b]

*p<0.05, **p<0.01, [†]Unadjusted OR

Abbreviations: NBS= Newborn screening; Info= information; OR= odds ratio; CI= confidence interval; HCP= Healthcare professional

[a] 230 respondents were excluded from these questions because they had indicated in a previous question (Question 10) that they either did not recall receiving information about newborn screening or that they did not know whether they received information.

[b] Odds ratios are compared to mothers who responded 'No' or 'Don't know' to questions about information received.

[c] The tear-off sheet is a double-sided page of information for parents that is attached to the blood collection card.

[d] All of these questions were separate so responses are not mutually exclusive (e.g.: mothers could report receiving information from both pre-natal class and the tear-off sheet).

Appendix 14: Table 37, Comparing study sample characteristics to those reported in the Canadian Maternity Experiences Survey

	NBS study, 2009	CMES, 2008
	% (95%CI)	%
Age (years)	N=748	N=6421
≤24	9 (7, 12)	18
25-29	24 (21, 27)	32
30-34	40 (37, 45)	33
≥35	27 (24, 31)	17
Education	N=746	N=6421
High school or less	15 (13, 18)	23
College	29 (25, 32)	43
University or higher		
Parity	N=749	N=6421
Primiparous	47 (43, 51)	45
Multiparous	53 (49, 57)	55

Abbreviations: NBS-newborn screening, CMES-Canadian Maternity Experiences Survey