Editor's key points

- Ongoing expansion of newborn screening (NBS) panels is resulting in increased numbers of infants with positive results, including carriers of genetic disorders and individuals with indeterminate results. This study explored cystic fibrosis (CF) as an exemplar of potential health system consequences of NBS expansion, examining primary care providers' preferred role and confidence in caring for infants receiving positive NBS results for CF.
- Overall, 77% of primary care providers indicated that they would not provide all well-baby care for infants with a confirmed diagnosis of CF, but most would share care with other specialists. Pediatricians were more disposed to provide care independently for infants with confirmed or inconclusive diagnoses, while midwives were less disposed to provide care independently.
- Primary care providers were significantly more likely to provide total well-baby or minor illness care for infants who were CF carriers if they were confident in reassuring parents about the health of infants who were carriers. Midwives were significantly less likely to provide total care, but are generally only involved in care in the first 6 weeks of life.

Newborn screening for cystic fibrosis

Role of primary care providers in caring for infants with positive screening results

June C. Carroll MD CCFP FCFP Robin Z. Hayeems ScM PhD Fiona A. Miller PhD Carolyn J. Barg MSc Yvonne Bombard PhD Pranesh Chakraborty MD FRCPC FCCMG Beth K. Potter PhD Jessica Peace Bytautas MA Karen Tam ScM Louise Taylor MN Elizabeth Kerr PhD Christine Davies MSc Jennifer Milburn MHA Felix Ratjen MD FRCPC Astrid Guttmann MSc MD CM

Abstract

Objective To explore primary care providers' (PCPs') preferred roles and confidence in caring for infants receiving a positive cystic fibrosis (CF) newborn screening (NBS) result, as well as management of CF family planning issues, given that expanded NBS has resulted in an increase in positive results.

Design Mailed questionnaire.

Setting Ontario.

Participants Ontario FPs, pediatricians, and midwives identified by Newborn Screening Ontario as having had an infant with a positive CF NBS result in their practice in the previous 6 months.

Main outcome measure Primary care providers' preferred roles in providing well-baby care for infants with positive CF screening results.

Results Overall, 321 of 628 (51%) completed surveys (208 FPs, 68 pediatricians, 45 midwives). For well-baby care for infants confirmed to have CF, 77% of PCPs indicated they would not provide total care (ie, 68% would share care with other specialists and 9% would refer to specialists completely); for infants with an inconclusive CF diagnosis, 50% of PCPs would provide total care, 45% would provide shared care, and 5% would refer to a specialist; for CF carriers, 89% of PCPs would provide total care, 9% would provide shared care, and 2% would refer. Half (54%) of PCPs were extremely or very confident in providing reassurance about CF carriers' health. Only 25% knew how to order parents' CF carrier testing; 67% knew how to refer for prenatal diagnosis. Confidence in reassuring parents about the health of CF carrier children was associated with providing total well-baby care for CF carriers (risk ratio of 1.50; 95% CI 1.14 to 1.97) and infants with an inconclusive diagnosis (risk ratio of 3.30; 95% CI 1.34 to 8.16).

Conclusion Most PCPs indicated willingness to treat infants with a range of CF NBS results in some capacity. It is concerning that some indicated CF carriers should have specialist involvement and only half were extremely or very confident about reassuring families about carrier status. This raises issues about possible medicalization of those with carrier status, prompting the need for PCP education about genetic disorders and the meaning of genetic test results.

Dépistage néonatal de la fibrose kystique

Le rôle des professionnels des soins primaires dans les soins aux nouveau-nés ont les résultats de dépistage sont positifs

June C. Carroll MD CCFP FCFP Robin Z. Hayeems ScM PhD Fiona A. Miller PhD Carolyn J. Barg MSc Yvonne Bombard PhD Pranesh Chakraborty MD FRCPC FCCMG Beth K. Potter PhD Jessica Peace Bytautas MA Karen Tam ScM Louise Taylor MN Elizabeth Kerr PhD Christine Davies MSc Jennifer Milburn MHA Felix Ratjen MD FRCPC Astrid Guttmann MSc MD CM

Résumé

Objectif Explorer les rôles que privilégient les professionnels des soins primaires (PSP) et leur confiance dans les soins aux nourrissons qui reçoivent des résultats positifs au dépistage néonatal (DN) de la fibrose kystique (FK), de même que dans la prise en charge des questions liées à la planification familiale pour la FK, étant donné que le DN élargi s'est traduit par une augmentation du nombre de résultats positifs.

Type d'étude Un questionnaire envoyé par la poste.

Contexte Ontario.

Participants Des MF, des pédiatres et des sages-femmes en Ontario, identifiés par Dépistage néonatal Ontario comme ayant pris soin d'un nouveau-né qui avait reçu des résultats positifs au DN de la FK durant les 6 mois précédents.

Principaux paramètres à l'étude Les rôles privilégiés par les PSP dans les soins de puériculture aux nouveau-nés ayant reçu des résultats positifs au dépistage de la FK.

Résultats Dans l'ensemble, 321 des 628 (51 %) PSP ont répondu au questionnaire (208 MF, 68 pédiatres, 45 sages-femmes). Pour les soins de puériculture aux nourrissons dont la FK était confirmée, 77 % des PSP ont indiqué qu'ils n'offriraient pas les soins complets (c.-à-d. que 68 % partageraient les soins avec d'autres spécialistes et 9 % s'en remettraient complètement à des spécialistes); pour les nourrissons dont le diagnostic de FK n'était pas concluant, 50 % offriraient les soins complets, 45 % fourniraient des soins partagés et 2 % aiguilleraient vers des soins spécialisés; pour les porteurs de la FK, 89 % des PSP offriraient les soins complets, 9 % des soins partagés, et 2 % aiguilleraient en spécialité. La moitié (54 %) des PSP avaient extrêmement ou très confiance de pouvoir rassurer les parents à propos de l'état de santé des porteurs de la FK. Seulement 25 % savaient comment prescrire un test pour les parents d'un porteur de la FK; 67 % savaient comment demander un diagnostic prénatal. La confiance de pouvoir rassurer les parents au sujet de la santé des enfants porteurs de la FK était associée à la prestation des soins de puériculture complets aux porteurs de la FK (risque relatif de 1,50; IC à 95 % de 1,14 à 1,97) et aux nourrissons dont le diagnostic n'était pas concluant (risque relatif de 3,30; IC à 95 % de 1,34 à 8,16).

Conclusion La plupart des PSP ont indiqué leur réceptivité à traiter dans une certaine mesure des nourrissons ayant reçu une gamme de résultats au DN de la FK. Il est préoccupant que certains aient indiqué que les porteurs de la FK devraient nécessiter l'implication de spécialistes et que seulement la moitié d'entre eux se sentaient extrêmement ou très confiants au sujet de la rassurance des familles concernant le fait d'être porteur. Cette situation soulève des questions entourant une médicalisation possible de ceux qui sont porteurs, et la nécessité d'une éducation des PSP au sujet des troubles génétiques et de la signification des résultats des tests génétiques.

Points de repère du rédacteur

- L'élargissement continuel des volets du dépistage néonatal (DN) se traduit par un plus grand nombre de nouveau-nés dont les résultats sont positifs, y compris des porteurs de troubles génétiques et d'autres dont les résultats sont indéterminés. Cette étude se penchait sur la fibrose kystique (FK) en guise d'exemple des conséquences potentielles dans le système de santé de l'expansion du DN, en examinant le rôle privilégié par les professionnels des soins primaires et leur confiance à s'occuper des nourrissons qui ont reçu des résultats positifs au DN de la FK.
- Dans l'ensemble, 77 % des professionnels des soins primaires ont indiqué qu'ils n'offriraient pas de soins de puériculture à des nourrissons ayant un diagnostic confirmé de FK, mais la plupart d'entre eux partageraient les soins avec d'autres spécialistes. Les pédiatres étaient plus disposés à prodiguer des soins de manière indépendante aux nourrissons ayant un diagnostic confirmé ou non concluant, tandis que les sages-femmes étaient moins réceptives à la prestation de soins de manière indépendante.
- Il était significativement plus probable que les professionnels des soins primaires fournissent tous les soins de puériculture ou pour des maladies mineures aux nourrissons porteurs de la FK s'ils avaient confiance de pouvoir rassurer les parents à propos de la santé des nourrissons porteurs. Les sages-femmes étaient significativement moins susceptibles de fournir l'ensemble des soins, mais elles ne sont généralement impliquées que durant les 6 premières semaines de vie.

Newborn screening (NBS) programs target serious treatable disorders that are not clinically evident in the newborn period. Newborn Screening Ontario (NSO) is the provincially funded program in Ontario that screens for rare diseases, including metabolic and endocrine diseases, sickle cell disease, cystic fibrosis (CF), severe combined immune deficiency, and spinal muscular atrophy, from the dried blood-spot sample collected at 1 to 2 days of age. In addition, there is bedside critical congenital heart disease screening through pulse oximetry.[1] Services are centralized and coordinated by NSO.

Ongoing expansion of NBS panels results in increased numbers of infants with positive results, including carriers of genetic disorders and individuals with indeterminate results. We studied CF as an exemplar of potential health system consequences of NBS expansion.

Cystic fibrosis is an inherited disease caused by mutations in the *CFTR* gene, resulting in impaired cellular electrolyte transfer. This causes abnormally thick mucus secretions in the respiratory, digestive, and reproductive systems.[2] Classic symptoms include lung infections and nutritional malabsorption. The incidence is about 1 in 2500 births in white populations[3] and 1 in 3600 in Canada.[4] Early diagnosis and treatment improve length and quality of life for people with CF, but life expectancy is shortened. Cystic fibrosis is autosomal recessive; 2 faulty copies of the *CFTR* gene are necessary to have the disease, 1 from each parent. Newborn screening identifies those with CF but also identifies some heterozygote carriers (1 copy of the faulty *CFTR* gene) and infants with nonclassic disease. In Ontario, NBS consists of screening for elevated immunoreactive trypsinogen (IRT), and if the IRT level is elevated, DNA *CFTR* mutation analysis is performed. Confirmatory sweat chloride testing follows if there is an elevated IRT level and 1 or 2 mutations are found. The final diagnosis can be confirmed CF (abnormal sweat test result), inconclusive screen-positive CF (an equivocal sweat test result), or CF carrier (normal sweat test result). Carriers do not have, and will not develop, CF. The importance is that 1 parent is usually also a CF carrier, and the couple, the infant later in life, and possibly extended family could be at risk of conceiving a child with CF.[5] Many studies have shown that parents report high anxiety or depression in reaction to positive CF NBS results, but most are relieved upon receiving normal sweat test results.[6-10]

In Ontario, infants receive care from publicly funded FPs, pediatricians (PEDs), or midwives (MWs). Midwives generally provide infant care for only the first 6 weeks of life. We are interested in how genetic testing, particularly NBS, affects the health care system, such as where and by whom care is provided. This is important as FPs and MWs might need to provide care for infants with positive NBS results, particularly in rural or remote areas where there are fewer PEDs. To this end, we explored care pathways for infants with positive screening results for CF on NBS, after confirmatory testing.

The objective of this study was to explore primary care providers' (PCPs') preferred roles in caring for infants who receive a positive CF NBS result and are confirmed to have CF, have an inconclusive CF diagnosis, or are CF carriers.

Methods

Study design

This is part of a longitudinal cohort study investigating the effects of CF NBS on families, health care providers, and health services in Ontario.[10-12] We received research ethics board approval from the University of Toronto in Ontario, the Children's Hospital of Eastern Ontario in Ottawa, and the Hospital for Sick Children in Toronto.

Sample and recruitment

We recruited PCPs (FPs, PEDs, MWs) identified by NSO as having had an infant with a positive CF NBS result in their practice in the previous 6 months. They were prospectively recruited between January 2012 and October 2013 (rolling, batched monthly) and mailed the survey package. Primary care providers were excluded if they did not practise in Ontario, or if the identified infant was younger than 33 weeks' gestation or deceased. The survey package included a self-complete questionnaire and a financial incentive (coffee shop coupon). We adapted the Dillman tailored design method: study notification letter and up to 3 reminder survey packages over 10 weeks. Completion and return of the questionnaire indicated consent to participate.

Data collection

We developed a structured, self-complete questionnaire that asked PCPs about their preferred role in providing routine well-baby care (eg, anticipatory guidance, administering vaccines, assessing growth and development) and care for minor acute illnesses (eg, upper respiratory tract infections, diarrhea) for infants confirmed to have CF, those with an inconclusive diagnosis of CF, and those who are carriers of CF. The questionnaire also assessed awareness of NBS conditions, whether participants believed they had an important role in NBS, their confidence in reassuring parents about the health of CF carriers and in discussing the relevance of carrier status for family planning, their ability to order CF carrier testing or refer for CF prenatal diagnosis, and demographic questions. Some questions were adapted from questionnaires used with similar study populations[13-21] or studies assessing similar PCP roles.[22-24] We pilot-tested the questionnaire with 15 PCPs. Midwives responded based on their care of infants during the first 6 weeks of life.

Analysis

Questionnaire data were entered into and analyzed using IBM SPSS, version 18. Double data entry was performed for 10% of the sample, with an error rate of less than 0.5%. We computed proportions, comparing across PCP groups using χ^2 and Fisher exact tests. Two-sided *P* values less than .05 indicated statistical significance. We looked at factors associated with PCPs providing total care versus providing shared and referred care for 4 clinical models.

- Model 1: well-baby care for infants who are CF carriers;
- Model 2: minor acute illness care for infants who are CF carriers;
- Model 3: well-baby care for infants who have an inconclusive CF diagnosis; and
- Model 4: minor acute illness care for infants who have an inconclusive CF diagnosis.

We used robust Poisson regression to analyze association of beliefs, confidence, and practice factors with self-reported approach to care of infants who were carriers or who had an inconclusive diagnosis. Informed by the literature, all variables were prespecified and entered into the adjusted regression model. All significant results are reported.

Results

Of 628 eligible PCPs, 321 (51%) completed surveys (208 FPs, 68 PEDs, 45 MWs). Demographic characteristics varied across provider type; however, most respondents were non-academic, in group practice, non–fee-for-service, urban, and female (**Table 1**).

For infants *confirmed* to have CF, most PCPs (77%) indicated they would not provide total well-baby care (**Table 2**). Most PCPs (68%) would share well-baby care with a specialist or subspecialist for infants *confirmed* to have CF (74% of FPs, 44% of PEDs, and 79% of MWs). Slightly more than half of PCPs (54%) would provide total care for minor illnesses for these infants (54% of FPs, 85% of PEDs, and 4% of MWs). For infants with an *inconclusive diagnosis* of CF, half of PCPs would provide total well-baby care (50% of FPs, 68% of PEDs, and 22% of MWs), and most (67%) would provide total care for minor illnesses (74% of FPs, 87% of PEDs, and 4% MWs). For infants who are CF *carriers,* 89% of PCPs would provide total well-baby care but 9% would share well-baby care with a specialist and 2% would refer totally to a specialist for well-baby care. By far most (84%) would provide total care to CF *carriers* for minor illnesses (97% of FPs, 94% of PEDs, and 9% of MWs), with 11% sharing care and 5% referring to specialists. Pediatricians were more disposed to provide care independently for infants with confirmed CF or inconclusive diagnoses, while MWs were less disposed to provide care independently for these children.

Slightly more than half the respondents (54%) were extremely or very confident in reassuring parents about the health of infants who were CF carriers (**Table 3**). Similarly, 59% were extremely or very confident in explaining the relevance of carrier status for family planning. Approximately 25% said they knew how to order CF carrier testing for adults, and 67% knew how to refer for prenatal diagnosis of CF.

Factors that were significant predictors of providing total care (well-baby or minor illness care) for infants who are CF *carriers* or have an *inconclusive* diagnosis of CF are shown in **Table 4**. Primary care providers were significantly more likely to provide total well-baby (risk ratio of 1.50; 95% CI 1.14 to 1.97) or minor illness care (risk ratio of 1.26; 95% CI 1.02 to 1.56) for infants who were CF *carriers* if they were extremely or very confident, or moderately confident, respectively, in reassuring parents about the health of infants who were CF carriers. They were significantly less likely to provide total care if they were MWs. There were no other consistent predictors of care across the 4 models.

Discussion

Most providers indicated willingness to provide care in some capacity for infants with screen-positive CF NBS results. Most PCPs would either share care with a specialist or provide total well-baby or minor illness care for infants with confirmed CF. Most PEDs indicated they would provide total care (well-baby and minor illness) for infants with confirmed CF, which is not surprising, given their training and expertise. Family physicians appeared to be more comfortable providing total well-baby and minor illness care for infants with an inconclusive diagnosis (50% and 74%, respectively) compared with infants with confirmed CF (17% and 54%, respectively). Some providers would share or refer for well-baby (11%) and minor illness (16%) care for infants who are carriers of CF—who are healthy infants. Only half of FPs (52%) and a third of MWs (31%) were very confident explaining the meaning of CF carrier status.

This study builds on literature showing that PCPs acknowledge a responsibility to integrate genetics into their practices,[25,26] but that they report limited knowledge.[25-27] Most NBS literature addresses PCPs' roles and challenges in notifying families about NBS results, with evidence of misconception about the conditions in NBS panels[13] and lack of perceived competence to discuss the meaning of positive screening results or confirmatory test results.[28] Studies have not addressed care of infants with confirmed, inconclusive, or carrier diagnoses. Specifically addressing CF, PCPs' lack of knowledge has been found in surveys regarding positive CF NBS results and carrier screening, including misperceptions about recessive inheritance.[29]

Midwives' responses might reflect guidelines that limit their scope of practice to caring for babies without abnormalities or disorders. Enabling midwifery care

Table 1. Sample characteristics

CHARACTERISTIC	FAMILY PHYSICIANS n (%), N = 208*	PEDIATRICIANS n (%), N = 68*	MIDWIVES n (%), N = 45	TOTAL n (%), N = 321*
Practice setting				
• Academic[†]	8 (3.8)	4 (5.9)	0 (0.0)	12 (3.7)
• Non-academic	200 (96.2)	64 (94.1)	45 (100.0)	309 (96.3)
Practice type				
• Solo	40 (19.2)	41 (61.2)	1 (2.2)	82 (25.6)
• Group[‡]	168 (80.8)	26 (38.8)	44 (97.8)	238 (74.4)
Method of reimbursement				
• Fee-for-service[§]	62 (29.8)	62 (91.2)	0 (0.0)	124 (38.6)
• Non–fee-for-service	146 (70.2)	6 (8.8)	45 (100.0)	197 (61.4)
Practice location				
• Urban	123 (59.4)	58 (85.3)	33 (73.3)	214 (66.9)
• Rural	84 (40.6)	10 (14.7)	12 (26.7)	106 (33.1)
Years in practice				
• 0-15	83 (39.9)	26 (38.2)	40 (88.9)	149 (46.4)
• ≥ 16	125 (60.1)	42 (61.8)	5 (11.1)	172 (53.6)
Sex				
• Female	130 (62.8)	28 (41.2)	45 (100.0)	203 (63.4)
• Male	77 (37.2)	40 (58.8)	0 (0.0)	117 (36.6)
Practice-based exposure to …				
• CF	58 (28.0)	37 (54.4)	10 (22.7)	105 (32.9)
• CF carrier	66 (32.2)	34 (52.3)	22 (51.2)	122 (39.0)
• NBS results positive for CF	47 (22.7)	33 (50.0)	26 (59.1)	106 (33.4)
• NBS results positive for another disorder	85 (41.1)	53 (80.3)	30 (69.8)	168 (53.2)

CF—cystic fibrosis, NBS—newborn screening.
*Denominators vary owing to missing data.
[†]Includes providers who see any patients in academic health sciences centres.
[‡]Includes interprofessional practice as well as single-discipline group practice.
[§]Includes providers who reported any fee-for-service. Midwives in Ontario are publicly funded but not fee-for-service.

in this context requires clarity that carrier infants do not have a disorder. As Ontario MWs care for infants for only the first 6 weeks following birth, they would be less involved in ongoing well-baby and minor illness care. Despite limits on scope and duration of involvement, being able to explain the meaning of CF carrier status is still important for MWs. A competency-based framework in genetics has been developed in the United Kingdom for midwifery education and practice, recognizing the role that MWs have in this area.[30]

The findings of this study have implications for both the NBS system and future genomic or personalized medicine tests that reveal carrier status, as some studies suggest that 2% of healthy individuals might be carriers of recessive pathogenic disease alleles.[31] There is a need for PCP education about the meaning of inconclusive and carrier results. Guidelines should be developed for addressing inconclusive NBS results, articulating how they should be interpreted and in what situations specialists or PCPs should be involved in care plans.[5] Care maps would be useful for patients who live in rural or remote areas who cannot readily access specialists or to determine when telemedicine might be a care alternative. Education of PCPs regarding carrier status is vital to improving confidence in providing care, patient education, and referring for family cascade testing and prenatal testing.[29]

Reviews of the psychological effects of infant carrier status on parents have shown that although most understand its benign implications, some retain misconceptions, have lingering anxiety about their children's health, and have longer-term concerns about stigma and reproductive implications.[2,5,8] It is important that PCPs communicate to parents that infants who are CF carriers are healthy and do not have CF.[5] Some studies have also shown unexpectedly high documented illness frequency among CF carrier infants[32] and higher

Table 2. Primary care providers' perspectives on their role in caring for children who have positive screening results for CF

ROLES	TOTAL n (%), N = 321*	FAMILY PHYSICIANS n (%), N = 208*	PEDIATRICIANS n (%), N = 68*	MIDWIVES n (%), N = 45*	*P* VALUE†
What role would you have in providing routine WBC for ...					
Infants confirmed to have CF					< .001
• Provide total WBC	73 (23.0)	35 (17.0)	37 (54.4)	1 (2.3)	
• Share WBC with specialist or subspecialist	216 (68.1)	152 (73.8)	30 (44.1)	34 (79.1)	
• Refer WBC to specialist or subspecialist	28 (8.8)	19 (9.2)	1 (1.5)	8 (18.6)	
Infants with an inconclusive diagnosis of CF					< .001
• Provide total WBC	158 (49.5)	102 (49.5)	46 (67.6)	10 (22.2)	
• Share WBC with specialist or subspecialist	144 (45.1)	99 (48.1)	21 (30.9)	24 (53.3)	
• Refer WBC to specialist or subspecialist	17 (5.3)	5 (2.4)	1 (1.5)	11 (24.4)	
Infants who are CF carriers					< .001
• Provide total WBC	284 (89.3)	196 (94.7)	61 (92.4)	27 (60.0)	
• Share WBC with specialist or subspecialist	27 (8.5)	11 (5.3)	1 (1.5)	15 (33.3)	
• Refer WBC to specialist or subspecialist	7 (2.2)	0 (0.0)	4 (6.1)	3 (6.7)	
What role would you have in providing care for minor acute illnesses for ...					
Infants confirmed to have CF					< .001
• Provide total care for minor acute illnesses	171 (53.6)	112 (54.1)	57 (85.1)	2 (4.4)	
• Share care for minor illnesses with specialist or subspecialist	113 (35.4)	84 (40.6)	10 (14.9)	19 (42.2)	
• Refer care for minor illnesses to specialist or subspecialist	35 (11.0)	11 (5.3)	0 (0.0)	24 (53.3)	
Infants with an inconclusive diagnosis of CF					< .001
• Provide total care for minor acute illnesses	214 (67.1)	154 (74.4)	58 (86.6)	2 (4.4)	
• Share care for minor illnesses with specialist or subspecialist	84 (26.3)	53 (25.6)	9 (13.4)	22 (48.9)	
• Refer care for minor illnesses to specialist or subspecialist	21 (6.6)	0 (0.0)	0 (0.0)	21 (46.7)	
Infants who are CF carriers					< .001
• Provide total care for minor illnesses	265 (83.6)	200 (96.6)	61 (93.8)	4 (8.9)	
• Share care for minor illnesses with specialist or subspecialist	36 (11.4)	7 (3.4)	1 (1.5)	28 (62.2)	
• Refer care for minor illnesses to specialist or subspecialist	16 (5.0)	0 (0.0)	3 (4.6)	13 (28.9)	

CF—cystic fibrosis, WBC—well-baby care.
*Denominators vary owing to missing data.
†Fisher exact test for difference in proportions across professions.

numbers of outpatient visits and hospitalization,[11] raising the question of heightened perceptions of vulnerability among healthy infants by parents and health care providers. This highlights the important PCP role in informing families about NBS results and thus the need to be familiar with NBS disorders and the meaning of false-positive and carrier test results.[33] Point-of-care information tools for providers and written and Web-based information for parents developed by various organizations[1] can help ensure that the meaning of results is well understood by both parties.[8,34]

Limitations

Respondents in this study might be more familiar with CF than average PCPs, as they had recently had an infant in their practice with a positive NBS result for CF. Responses reflect intention to provide care not actual care delivery.

Table 3. Primary care providers' perspectives and confidence related to NBS and CF carrier status

FACTOR	TOTAL n (%), N = 321*	FAMILY PHYSICIANS n (%), N = 208*	PEDIATRICIANS n (%), N = 68*	MIDWIVES n (%), N = 45*	*P* VALUE
Up to date on NBS					<.001†
• Strongly agree or agree	182 (57.6)	90 (43.7)	57 (85.1)	35 (81.4)	
• Strongly disagree, disagree, or neutral	134 (42.4)	116 (56.3)	10 (14.9)	8 (18.6)	
Important role in NBS					<.001†
• Strongly agree or agree	256 (80.8)	153 (74.3)	59 (88.1)	44 (100.0)	
• Strongly disagree, disagree, or neutral	61 (19.2)	53 (25.7)	8 (11.9)	0 (0.0)	
Confidence in providing reassurance about health of CF carrier					<.001†
• Extremely or very confident	172 (53.9)	107 (51.7)	51 (76.1)	14 (31.1)	
• Moderately, not very, or not at all confident	147 (46.1)	100 (48.3)	16 (23.9)	31 (68.9)	
Within scope to provide reassurance about health of CF carrier					.001†
• Yes	268 (88.2)	176 (88.9)	60 (96.8)	32 (72.7)	
• No	36 (11.8)	22 (11.1)	2 (3.2)	12 (27.3)	
Confidence in explaining relevance of carrier status to family planning					.009‡
• Extremely or very confident	187 (58.6)	112 (54.1)	52 (77.6)	23 (51.1)	
• Moderately confident	109 (34.2)	78 (37.7)	13 (19.4)	18 (40.0)	
• Not very or not at all confident	23 (7.2)	17 (8.2)	2 (3.0)	4 (8.9)	
Within scope to explain relevance of carrier status					.39†
• Yes	269 (84.6)	177 (85.9)	57 (85.1)	35 (77.8)	
• No	49 (15.4)	29 (14.1)	10 (14.9)	10 (22.2)	
Know how to order CF carrier testing for adults					.006†
• Yes	79 (24.9)	60 (29.4)	16 (23.5)	3 (6.7)	
• No	238 (75.1)	144 (70.6)	52 (76.5)	42 (93.3)	
Within scope to order CF carrier testing for adults					<.001†
• Yes	150 (47.9)	132 (65.7)	15 (22.4)	3 (6.7)	
• No	163 (52.1)	69 (34.3)	52 (77.6)	42 (93.3)	
Know how to refer for prenatal diagnosis related to CF					<.001†
• Yes	213 (67.0)	132 (64.4)	39 (57.4)	42 (93.3)	
• No	105 (33.0)	73 (35.6)	29 (42.6)	3 (6.7)	
Within scope to refer for prenatal diagnosis related to CF					<.001†
• Yes	257 (81.1)	176 (85.9)	37 (55.2)	44 (97.8)	
• No	60 (18.9)	29 (14.1)	30 (44.8)	1 (2.2)	

CF—cystic fibrosis, NBS—newborn screening.
*Denominators vary owing to missing data.
†χ^2 test.
‡Fisher exact test.

Conclusion

Few studies have examined PCPs' preferred practices with infants with CF or carriers of genetic disorders. This study found that most PCPs are willing to care for infants with a range of screen-positive CF NBS results in some capacity. However, providers' lack of confidence to reassure about carrier status raises issues about its possible medicalization, prompting the need for specific

Table 4. Factors associated with PCPs providing total care vs shared and referred care

FACTOR	MODEL 1: PCP PROVIDING TOTAL* WBC FOR INFANTS WHO ARE CF CARRIERS, ADJUSTED RR (95% CI)	MODEL 2: PCP PROVIDING TOTAL* ACUTE MINOR ILLNESS CARE FOR INFANTS WHO ARE CF CARRIERS, ADJUSTED RR (95% CI)	MODEL 3: PCP PROVIDING TOTAL* WBC FOR INFANTS WHO HAVE AN INCONCLUSIVE DIAGNOSIS OF CF, ADJUSTED RR (95% CI)	MODEL 4: PCP PROVIDING TOTAL* ACUTE MINOR ILLNESS CARE FOR INFANTS WHO HAVE AN INCONCLUSIVE DIAGNOSIS OF CF, ADJUSTED RR (95% CI)
Extremely or very confident in reassuring about health of CF carrier vs not very or not at all confident	1.50 (1.14-1.97)†	1.21 (0.98-1.50)	3.30 (1.34-8.16)†	1.61 (1.03-2.52)†
Moderately confident in reassuring about health of CF carrier vs not very or not at all confident	1.44 (1.10-1.90)†	1.26 (1.02-1.56)†	4.08 (1.68-9.92)†	1.89 (1.21-2.95)†
Population served: urban vs rural	0.92 (0.86-0.99)†	0.95 (0.89-1.02)	0.93 (0.72-1.19)	0.94 (0.81-1.10)
Female vs male	1.16 (1.07-1.26)†	1.08 (1.00-1.16)†	1.06 (0.84-1.33)	0.92 (0.80-1.06)
Midwives vs family physicians and pediatricians‡	0.72 (0.56-0.94)†	0.12 (0.05-0.31)§	0.46 (0.22-0.98)†	0.08 (0.02-0.32)§
Within scope to provide reassurance about health of CF carrier: yes vs no	1.17 (0.94-1.47)	1.02 (0.90-1.14)	0.64 (0.43-0.97)†	0.89 (0.69-1.16)
Child in practice with positive NBS result for CF: yes vs no	1.05 (0.95-1.17)	1.06 (0.97-1.15)	1.09 (0.82-1.44)	1.32 (1.12-1.55)†

CF—cystic fibrosis, NBS—newborn screening, PCP—primary care provider, RR—risk ratio, WBC—well-baby care.
*Total care vs combined shared and referred care.
†$P < .05$.
‡Differences between family physicians vs midwives and pediatricians, and pediatricians vs midwives and family physicians were not significant.
§$P < .01$.

PCP education about genetic disorders and the meaning of genetic test results, particularly carrier status. This will become increasingly important as genome-wide sequencing strategies enter NBS algorithms.[35] Education and care guidelines for children with CF or inconclusive CF diagnoses would enable PCPs to provide well-baby and minor illness care with more confidence, particularly in rural and remote areas.

Dr Carroll is a family physician and clinician scientist, Professor, and Sydney G. Frankfort Chair in Family Medicine in the Department of Family and Community Medicine with the Sinai Health System and the University of Toronto in Ontario. **Dr Hayeems** is a scientist in the Child Health Evaluative Sciences Program at the Hospital for Sick Children in Toronto and Associate Professor in the Institute of Health Policy, Management and Evaluation at the University of Toronto. **Dr Miller** is Professor of Health Policy and holds the Chair in Health Management Strategies at the Institute of Health Policy, Management and Evaluation at the University of Toronto. **Ms Barg** is a research analyst at Health Quality Ontario in Toronto. **Dr Bombard** is a scientist at the Li Ka Shing Knowledge Institute at St Michael's Hospital in Toronto and Associate Professor in the Institute of Health Policy, Management and Evaluation at the University of Toronto. **Dr Chakraborty** is Executive Director and Chief Medical Officer of Newborn Screening Ontario in Ottawa, a medical biochemist in the Department of Pediatrics at the Children's Hospital of Eastern Ontario in Ottawa and Associate Professor of Pediatrics at the University of Ottawa. **Dr Potter** is Associate Professor in the Department of Epidemiology and Community Medicine at the University of Ottawa and holds the University Research Chair in Health Services for Children with Rare Diseases. **Ms Bytautas** is a doctoral student in the Dalla Lana School of Public Health and a research assistant in the Institute of Health Policy, Management and Evaluation at the University of Toronto. **Ms Tam** is a certified genetic counselor and screening specialist at Newborn Screening Ontario. **Ms Taylor** is a nurse practitioner with expertise in caring for children with cystic fibrosis at the Hospital for Sick Children. **Dr Kerr** is a clinical neuropsychologist and a scientist in the Department of Pediatrics, Division of Neurology, at the Hospital for Sick Children, and Adjunct Faculty in the Department of Pediatrics at the University of Toronto. **Ms Davies** is a certified genetic counselor in Ottawa. **Ms Milburn** is Operations Director for Newborn Screening Ontario. **Dr Ratjen** is Division Chief of Pediatric Respiratory Medicine, Co-lead of the Cystic Fibrosis Centre, Senior Scientist at the Research Institute in the Translational Medicine research program, and Medical Director of the Clinical Research Unit, all at the Hospital for Sick Children, and Professor of Pediatrics at the University of Toronto. **Dr Guttmann** is a clinician scientist in the Division of Pediatric Medicine at the Hospital for Sick Children, Chief Science Officer and Senior Scientist at ICES, and Professor of Pediatrics with a cross appointment at the Institute of Health Policy, Management and Evaluation and the Epidemiology Division of the Dalla Lana School of Public Health at the University of Toronto.

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Contributors
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Competing interests
None declared

Correspondence
Dr June C. Carroll; e-mail **june.carroll@sinaihealth.ca**

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