Primary care providers' role in newborn screening result notification for cystic fibrosis

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Abstract

Objective To explore primary care providers' (PCPs') role in result notification for newborn screening (NBS) for cystic fibrosis (CF), given that expanded NBS has increased the number of positive screening test results, drawing attention to the role of PCPs in supporting families.

Design Cross-sectional survey and qualitative interviews.

Setting Ontario.

Participants Primary care providers (FPs, pediatricians, and midwives) who received a positive CF NBS result for an infant in their practice in the 6 months before the study.

Main outcome measures Whether the PCP notified the family of the initial positive CF screening result.

Results Data from 321 PCP surveys (response rate of 51%) are reported, including 208 FPs, 68 pediatricians, and 45 midwives. Interviews were completed with 34 PCPs. Most (65%) surveyed PCPs reported notifying the infant's family of the initial positive screening result; 81% agreed that they have an important role to play in NBS; and 88% said it was important for PCPs, rather than the NBS centre, to notify families of initial positive results. With support and information from NBS centres, 68% would be extremely or very confident in doing so; this dropped to 54% when reflecting on their recent reporting experience. More than half (58%) of all PCPs said written point-of-care information from the NBS centre was the most helpful format. Adjusted for relevant factors, written educational information was associated with a lower rate of notifying families than written plus verbal information (risk ratio of 0.79; 95% CI 0.69 to 0.92). In the interviews, PCPs emphasized the challenge of balancing required content knowledge with the desire for the news to come from a familiar provider.

Conclusion Most PCPs notify families of NBS results and value this role. These data are relevant as NBS programs and other genomic services expand and consider ways of keeping PCPs confident and actively involved.

Editor's key points

- New technologies and targets are being implemented into newborn screening programs, increasing the importance of primary care provider involvement in supporting this population-wide program.
- Primary care providers endorse the importance of their involvement in notification of positive results and value point-of-care information from newborn screening programs to enable effective communication with families in their care.
- Continued effort to engage primary care providers in genomic medicine is warranted.

Points de repère du rédacteur

- Les programmes de dépistage néonatal utilisent de nouvelles technologies et se fixent d'autres cibles, ce qui rehausse l'importance de la participation des professionnels des soins primaires dans le soutien à ces programmes populationnels.
- Les professionnels des soins primaires conviennent de l'importance de leur implication dans la révélation des résultats positifs, et ils valorisent l'obtention de renseignements au point de service de la part des programmes de dépistage néonatal pour permettre une communication efficace avec les familles qu'ils soignent.
- Il est justifié de poursuivre les efforts de mobilisation des professionnels des soins primaires dans la médecine génomique.

Le rôle des professionnels des soins primaires dans la communication des résultats du dépistage néonatal de la fibrose kystique

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Résumé

Objectif Explorer le rôle des professionnels des soins primaires (PSP) dans la communication des résultats du dépistage néonatal (DN) de la fibrose kystique (FK), étant donné que le DN élargi a augmenté le nombre de résultats de tests de dépistage positifs et a attiré l'attention sur le rôle des PSP dans le soutien aux familles.

Type d'étude Sondage transversal et entrevues qualitatives.

Contexte Ontario.

Participants Les professionnels des soins primaires (MF, pédiatres et sages-femmes) qui ont reçu des résultats de DN de la FK pour un nouveau-né dans leur pratique durant les 6 mois précédant l'étude.

Principaux paramètres à l'étude La communication à la famille par le PSP des résultats positifs initiaux du dépistage de la FK.

Résultats Les données tirées de sondages auprès de 321 PSP (taux de réponse de 51%) sont rapportées, venant notamment de 208 MF, 68 pédiatres et 45 sages-femmes. Des entrevues ont été effectuées avec 34 PSP. La plupart (65%) des PSP qui ont répondu au sondage ont signalé avoir communiqué les résultats positifs initiaux à la famille du nouveau-né; 81% ont convenu qu'ils avaient un rôle important à jouer dans le DN; et 88% ont dit qu'il était très important que les PSP, plutôt que les centres de DN, avertissent les familles des résultats positifs initiaux. S'ils disposaient du soutien et de renseignements des centres de DN, 68% seraient extrêmement ou très à l'aise de le faire; ce pourcentage a baissé à 50% lorsque les PSP se sont rappelé leur récente expérience de communication. Environ la moitié (52%) de tous les PSP ont indiqué que des renseignements par écrit au point de service étaient la forme la plus utile. Après rajustement en fonction des facteurs pertinents, des renseignements éducatifs par écrit étaient associés à un taux plus faible de communication avec les familles que de l'information écrite accompagnée par des renseignements verbaux (risque relatif de 0,79; IC à 95% de 0,69 à 0,92). Dans les entrevues, les PSP ont insisté sur le défi que représente un juste équilibre entre les connaissances requises du sujet et le désir que les résultats soient communiqués par un professionnel connu de la famille.

Conclusion La plupart des PSP communiquent aux familles les résultats du DN et valorisent ce rôle. Ces données sont d'autant plus pertinentes que les programmes de DN et d'autres services génomiques prennent de l'expansion et envisagent des façons de préserver la confiance des PSP et leur implication active.

Novel genomic technologies promise to mitigate disease burden through early detection and targeted intervention.[1] Integrating primary care providers (PCPs) into the systems of care that deliver these technologies is integral to their success. However, efforts to do so have identified gaps in content knowledge and confidence, time constraints, limited access to genetics experts, referral guidelines, and point-of-care tools, and poor integration of laboratory results into electronic medical records.[2]

Newborn screening (NBS), the *heel prick test*, presents a context that increases both the opportunity and the need for timely involvement of PCPs.[3,4] The growing number and increasing complexity of positive NBS results—particularly in light of screening algorithms that will soon use next-generation sequencing as a first-tier test and biochemistry as a second-tier test[5]—highlights the importance of maintaining PCP involvement in NBS care (eg, of 1750 screen-positive results in Ontario annually, 11% are true positive, 57% are false positive, 20% are carriers, and 5% are secondary disease targets).[6] Given the range of results and the ongoing expansion of screening programs, an emphasis on the practical effects of these findings for PCPs is warranted. Related work has emphasized the importance of the PCPs' postnatal role in educating and communicating with families about NBS results[3,7-11] and the importance of ongoing capacity building among PCPs to maintain integrated care in this context.

Newborn screening for cystic fibrosis (CF)—using the immunoreactive trypsinogen assay followed by DNA mutation analysis—is a helpful paradigm for examining the NBS system, as it represents the largest single-disease contributor to the total number of infants with positive screening results (30%).[12] Herein we describe Ontario providers' actual experience notifying families of positive screening results and determine the factors associated with their involvement in this component of care.

Methods

Design and setting

This study draws from a larger cohort study that evaluated NBS for CF in Ontario.[13-15] We received research ethics 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. Newborn Screening Ontario (NSO) is a centralized provincial program that screens approximately 140 000 infants annually. When a screen-positive result is identified, a representative from 1 of 5 regional centres attempts to contact the PCP on record. If the NSO representative reaches the PCP on record and this provider chooses to notify the family, the retrieval centre provides written- and telephone-based point-of-care information to the PCP.[16] If this PCP cannot be reached or chooses to delegate notification, the regional centre notifies the family.

Sample and recruitment

Prospectively, we recruited PCPs (FPs, pediatricians [PEDs], and midwives [MWs]) who were verified to be the provider on record for an infant with positive CF screening results in 2012 to 2013. Primary care providers were excluded if they did not practise in Ontario, if contact information was incomplete, or if the index infant was younger than 33 weeks' gestation, deceased, or in the care of the Children's Aid Society. The survey package included a self-complete questionnaire, an information sheet, and a coffee shop coupon. Following the Dillman method, we contacted potential participants by mail with a study notification letter and up to 3 reminders, over a 10-week period. Completing the questionnaire constituted consent. Respondents were also invited to participate in a qualitative interview, for which consent was obtained.

Data collection

Our structured questionnaire assessed providers' awareness of NBS, perspectives on the PCP role in result notification and care of CF carrier infants, and thoughts on modifiable barriers to PCP involvement. Our survey instrument was adapted from questionnaires used with similar study populations[10,17-24] or that addressed similar PCP dynamics[25-27] and was pilot-tested with a convenience sample of PCPs in Toronto (N=15). Two members of the study team (J.P.B. and S.P.) conducted qualitative telephone interviews. The interview guide queried providers' engagement with NBS, perceived and desired roles, experiences, and barriers related to notifying families of positive screening results.

Analysis

Questionnaire data were manually entered into IBM SPSS, version 18. Double data entry was performed for 10% of the sample but not completed for the full data set (error rate < 0.5%). We quantified the overall pattern of responses, developing frequency distributions, comparing across PCP groups. We used χ^2 and Fisher exact tests; 2-sided P values less than .05 indicated statistical significance. We used robust Poisson regression to analyze the association of beliefs, confidence, and practice factors with actual notification of positive screening results, controlling for PCP characteristics. All variables were prespecified and entered into the adjusted regression model.

Interviews were audiorecorded, transcribed, and coded thematically. Both deductive and inductive codes were generated independently by the authors (R.Z.H. and F.A.M.), discussed, and agreed upon. Transcripts were then independently coded by an additional author (S.P.). Themes were established and refined through an iterative process.

Results

Participants

Of 628 eligible PCPs, 321 completed surveys (51%). Overall, 65% of respondents were FPs, 21% were PEDs,

and 14% were MWs. Before this study, 33% to 39% had had exposure to a patient with CF, a CF carrier, or an infant with a positive CF screening result, and 53% had been exposed to a patient with a positive NBS result for another condition. Compared with PEDs and MWs, FPs reported the least exposure to these scenarios (23% to 41%; **Table 1**). Qualitative interviews were completed by 34 respondents; they had less practice-based exposure to CF, but similar levels of exposure to CF carriers or positive NBS screening results (**Table 2**).

Survey findings

Perspectives on NBS (Table 3). While more than 80% of PEDs and MWs reported being up to date about NBS, only 44% of FPs reported this. By far, most perceived that they had an important role to play in result notification (88% of FPs, 84% of PEDs, and 98% of MWs); however, anticipated confidence related to reporting results was lower (64% of FPs, 75% of PEDs, and 80% of MWs), and this dropped to 54% overall when PCPs reflected on their recent reporting experience. While providers did not expect to face practice-based barriers to reporting NBS results within 3 days (72% of FPs, 57% of PEDs, and 82% of MWs), most PEDs and FPs expected to face at least moderate barriers to reporting (more urgent) results within 1 day (67% of FPs, 68% of PEDs, and 41% of MWs). Pediatricians anticipated more barriers to notification than other groups did. Primary care providers reported a preference for both verbal and written point-of-care information (59% of FPs, 71% of PEDs, and 86% of MWs).

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.

Table 2. Sample characteristics of interview respondents

CHARACTERISTIC	FAMILY PHYSICIANS n (%), N = 17	PEDIATRICIANS n (%), N = 4	MIDWIVES n (%), N = 13*	TOTAL n (%), N = 34
Practice setting				
• Academic†	0 (0.0)	1 (25.0)	0 (0.0)	1 (2.9)
• Non-academic	17 (100.0)	3 (75.0)	13 (100.0)	33 (97.1)
Practice type				
• Solo	1 (5.9)	0 (0.0)	1 (7.7)	2 (5.9)
• Group‡	16 (94.1)	4 (100.0)	12 (92.3)	32 (94.1)
Method of reimbursement				
• Fee-for-service§	4 (23.5)	3 (75.0)	0 (0.0)	7 (20.6)
• Non–fee-for-service	13 (76.5)	1 (25.0)	13 (100.0)	27 (79.4)
Practice location				
• Urban	9 (52.9)	4 (100.0)	10 (76.9)	23 (67.6)
• Rural	8 (47.1)	0 (0.0)	3 (23.1)	11 (32.4)
Years in practice				
• 0-15	13 (76.5)	3 (75.0)	11 (84.6)	27 (79.4)
• ≥ 16	4 (23.5)	1 (25.0)	2 (15.4)	7 (20.6)
Sex				
• Female	14 (82.4)	4 (100.0)	13 (100.0)	31 (91.2)
• Male	3 (17.6)	0 (0.0)	0 (0.0)	3 (8.8)
Practice-based exposure to …				
• CF	4 (23.5)	2 (50.0)	3 (23.1)	9 (26.5)
• CF carrier	6 (35.3)	1 (25.0)	6 (46.2)	13 (38.2)
• NBS results positive for CF	4 (23.5)	2 (50.0)	7 (53.8)	13 (38.2)
• NBS results positive for another disorder	5 (29.4)	4 (100.0)	8 (61.5)	17 (50.0)

CF—cystic fibrosis, NBS—newborn screening.
*Fifteen midwives were interviewed but the audio for 1 interview (with 2 midwives) did not record properly, so data are reported for 13 participants.
†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.

Actual experience with NBS (Table 4). Most FPs and MWs had pre-existing relationships with index families, whereas only 43% of PEDs did. When given the opportunity to notify families of NBS results, most reported doing so (67% of FPs, 72% of PEDs, and 77% of MWs). Among FPs, 46% reported feeling confident with notification, whereas 74% of PEDs and 64% of MWs did. Among those who did not notify families themselves, "reported by NSO" was the most frequent reason (77%; data not shown). In practice, written point-of-care information was received more often than verbal information was by FPs (55% vs 18%) and PEDs (71% vs 20%) and was more highly valued by FPs (60% vs 28%) and PEDs (59% vs 16%) compared with MWs (46% vs 50%).

Factors associated with notification (Table 5). An in-principle commitment to NBS trended toward an association with actual result notification in practice (risk ratio of 1.31; 95% CI 1.00 to 1.71). Written information alone from the NBS centre was associated with a lower rate of notifying families than written plus verbal information (risk ratio of 0.79; 95% CI 0.69 to 0.92).

Interview findings

In the interviews, many respondents emphasized how patients value information coming from a known PCP, while others deferred notification to the program, doubting their content expertise. For most providers, the existing integrated system design—in which NSO acts as liaison for the provider, equips them with essential

Table 3. Primary care providers' perspectives on NBS and notification of results

CHARACTERISTIC	FAMILY PHYSICIANS n (%), N = 208*	PEDIATRICIANS n (%), N = 68*	MIDWIVES n (%), N = 45*	*P* VALUE
Up to date on NBS				< .01†
• Strongly agree or agree	90 (43.7)	57 (85.1)	35 (81.4)	
• Strongly disagree, disagree, or neutral	116 (56.3)	10 (14.7)	8 (18.6)	
Important role in NBS in general				< .01†
• Strongly agree or agree	153 (74.3)	59 (88.1)	44 (100.0)	
• Strongly disagree, disagree, or neutral	53 (25.7)	8 (11.9)	0 (0.0)	
Important role in notification of NBS results				.07†
• Strongly agree or agree	183 (88.4)	56 (83.6)	44 (97.8)	
• Strongly disagree, disagree, or neutral	24 (11.6)	11 (16.4)	1 (2.2)	
Confidence regarding notification				.17‡
• Extremely or very confident	132 (63.8)	51 (75.0)	36 (80.0)	
• Moderately confident	57 (27.5)	14 (20.6)	8 (17.8)	
• Not very or not at all confident	18 (8.7)	3 (4.4)	1 (2.2)	
Perceived barriers to notifying within 3 d				.02‡
• Extremely or very significant	17 (8.2)	7 (10.3)	0 (0.0)	
• Moderately significant	40 (19.3)	22 (32.4)	8 (17.8)	
• Not very or not at all significant	150 (72.5)	39 (57.4)	37 (82.2)	
Perceived barriers to notify within 1 d				.01†
• Extremely or very significant	72 (35.0)	28 (43.1)	9 (20.5)	
• Moderately significant	65 (31.6)	16 (24.6)	9 (20.5)	
• Not very or not at all significant	69 (33.5)	21 (32.3)	26 (59.1)	
Preference for point-of-care information§				< .01‡
• Written information	44 (21.7)	13 (19.7)	6 (13.6)	
• Verbal communication with treatment centre provider	17 (8.4)	2 (3.0)	0 (0.0)	
• Both written and verbal	120 (59.1)	47 (71.2)	38 (86.4)	
• Either written or verbal (does not matter which)	22 (10.8)	4 (6.1)	0 (0.0)	

NBS—newborn screening.
*Denominators vary owing to missing data.
†χ^2 test.
‡Fisher exact test.
§Excluded 7 respondents who selected more than 1 response and 1 who selected "other."

point-of-care facts, and determines the best notification plan—is working well. However, some reflected upon system fallibility when, for example, "I get a panicked call … and I'm scrambling trying to get information before I call [Dad] back, in the middle of clinic" (**Table 6**).

Discussion

Our findings show that most PCPs agreed that they have an important role in NBS and, specifically, an important role in notification of positive screening results. For the recent infant in their care, most reported having notified and discussed positive results with the family. This was largely driven by a principled belief in maintaining a role in the screening system. Qualitative data show that providers believe that patients value notification from a familiar professional, despite concerns about their own content expertise, lending further support to the importance of their role. The value of the information provided by the screening program was emphasized as mitigating these concerns. While theoretically providers preferred receiving written and verbal information, most received only written information during their actual notification experience and valued it highly.

We identified variation across PCP groups enrolled in our study, highlighting specific characteristics and conditions that might warrant further attention. For example, while more than 80% of PEDs and MWs reported being up to date on NBS, only 44% of FPs shared this view. In previous work (2009),[20] soon after NBS expanded in Ontario, 46% of FPs reported feeling up to date. Suboptimal awareness among FPs—particularly when

Table 4. Primary care providers' actual experiences with NBS result notification

EXPERIENCE	FAMILY PHYSICIANS n (%), N = 163*	PEDIATRICIANS n (%), N = 45*	MIDWIVES n (%), N = 35*	*P* VALUE
Notified family				.46†
• Yes	103 (66.9)	28 (71.8)	27 (77.1)	
• No	51 (33.1)	11 (28.2)	8 (22.9)	
Confidence when notifying family				.03‡
• Extremely or very confident	56 (45.5)	25 (73.5)	18 (64.3)	
• Moderately confident	51 (41.5)	8 (23.5)	9 (32.1)	
• Not very or not at all confident	16 (13.0)	1 (2.9)	1 (3.6)	
Point-of-care information received				< .01†
• Written information	67 (54.9)	25 (71.4)	8 (28.6)	
• Verbal communication with treatment centre provider	22 (18.0)	7 (20.0)	8 (28.6)	
• Both written and verbal	33 (27.0)	3 (8.6)	12 (42.9)	
Most helpful point-of-care information§				.02‡
• Written information from NBS program	67 (59.8)	19 (59.4)	11 (45.8)	
• Verbal information from NBS program	31 (27.7)	5 (15.6)	12 (50.0)	
• Own prior knowledge	9 (8.0)	8 (25.0)	1 (4.2)	
• Other	5 (4.5)	0 (0.0)	0 (0.0)	
Existing relationship with family				< .01†
• Yes	140 (92.1)	17 (42.5)	32 (91.4)	
• No	12 (7.9)	23 (57.5)	3 (8.6)	
Caring for infant since birth				.40‡
• Yes	145 (94.8)	36 (90.0)	32 (91.4)	
• No	8 (5.2)	4 (10.0)	3 (8.6)	

NBS—newborn screening, NSO—Newborn Screening Ontario.
*No. of respondents who reported being contacted by NSO about screen-positive results for a child in their practice. Denominators vary owing to missing data.
†χ^2 test.
‡Fisher exact test.
§Excludes 17 respondents who checked more than 1 option when asked to select 1.

sustained over time—warrants consideration by both the NBS and FP communities. This might be a partial explanation for the finding that FPs were less likely to perceive an important role for themselves and were less confident about result notification than PEDs and MWs were. Qualitative reservations about content expertise align with these findings. While seemingly not as prepared for the task as they might have preferred to be (or perhaps more humble in their reporting), FPs' belief in their role, access to effective point-of-care tools, and engagement with the families in their care appear to have motivated proactive involvement. Literature that reflects on the role of PCPs in post-NBS care is limited and focuses primarily on communication practices and quality.[3,7-11] In addition, it is largely drawn from non-Canadian settings where the links between the local screening programs and PCPs are not as well established as they are in Canada. For this reason, our data help to reflect on specific provider characteristics, practice conditions, and qualitative experiences that are associated with PCP engagement in post-NBS care.

Our findings can inform ongoing efforts to optimize PCPs' roles in NBS and other types of genomic screening and diagnostics. While our findings reflect on the specific context of NBS for CF, we speculate that providers' perceived role, commitment to families, and active use of point-of-care tools will be essential to their sustained integration into delivering genomic medicine. Related work conducted by Carroll et al[2] identified that strong ties to local genetics services are also essential to this integration. Similarly, ongoing efforts to optimize electronic interfaces, communication processes, and continuing education between PCPs and centralized genomic screening programs should be maintained. In our view, these core enablers will hold true as testing technologies and algorithms evolve. While emerging screening algorithms for CF are expected to reduce the rate of false positives,[28] PCPs will be the recipients of positive results related to novel targets, but are already central to increasingly genomics-driven prenatal screening[29] and might soon play a central role in navigating the application of genome-wide sequencing to their practices.[30,31]

Table 5. Factors associated with PCP notification of parents of children with positive CF NBS results among those contacted by NSO

FACTOR	ADJUSTED RR (95% CI)
Beliefs	
Beliefs about PCP role in NBS	
• Important role in general (agree vs disagree)	1.31 (1.00-1.71)*
• Important role in notification (agree vs disagree)	1.25 (0.83-1.88)
Confidence	
Confident making the notification	
• Extremely or very confident vs not very or not at all confident	1.26 (0.76-2.09)
• Moderately confident vs not very or not at all confident	1.05 (0.61-1.79)
Preparedness	
Point-of-care information received	
• Verbal communication with treatment centre vs both written and verbal	0.96 (0.86-1.08)
• Written information vs both written and verbal	0.79 (0.69-0.92)†
Practice-based exposure to …	
• CF (yes vs no)	1.11 (0.98-1.26)
• CF carrier (yes vs no)	0.95 (0.84-1.08)
• NBS results positive for CF (yes vs no)	1.01 (0.87-1.18)
• NBS results positive for another disorder (yes vs no)	1.03 (0.89-1.20)
Relationship with family	
Existing relationship with family (yes vs no)	0.87 (0.73-1.04)
Caring for infant since birth (yes vs no)	1.06 (0.75-1.50)
Perceived barriers	
Practice-related barriers to notify within 1 d	
• Extremely or very significant vs not very or not at all significant	1.11 (0.91-1.36)
• Moderately significant vs not very or not at all significant	1.07 (0.91-1.24)
Practice-related barriers to notify within 3 d	
• Extremely or very significant vs not very or not at all significant	0.97 (0.71-1.31)
• Moderately significant vs not very or not at all significant	0.86 (0.68-1.09)
PCP characteristics	
Practice type (solo vs group)	0.96 (0.80-1.16)
Practice location (urban vs rural)	0.97 (0.85-1.09)
Years in practice (0-15 vs ≥16)	1.07 (0.93-1.22)
Sex (female vs male)	1.07 (0.93-1.22)
Provider type	
• Family physician vs midwife	1.04 (0.85-1.27)
• Pediatrician vs midwife	0.96 (0.73-1.26)

CF—cystic fibrosis, NBS—newborn screening, NSO—Newborn Screening Ontario, PCP—primary care practitioner, RR—risk ratio.
*$P < .05$.
†$P < .01$.

Limitations

Our response rate was low and respondents represent the experience of those who are more invested in NBS. Data are also limited by possible recall bias. As our data reflect the experience of Ontario providers, its applicability to jurisdictions where NBS programs interface differently with PCPs might be limited. However, on comparable characteristics ascertained by the National Physician Survey, our sample is aligned (eg, <10% in academic centres, <40% in practice for 0 to 15 years).[32] While data herein address result notification, the survey also ascertained PCPs' role in post-NBS care (**page e144**).[33] Finally, while these data were collected in 2012 and respondents' views might differ today, PCPs' knowledge, attitudes, and confidence related to genetics tends to remain stable over time.[34]

Table 6. Qualitative findings from interviews with PCPs: *N* = 34.

CORE THEME	SAMPLE DATA REPRESENTING EACH THEME
Balancing pre-existing relationship with required expertise	"OK, so a stranger calls you from the hospital and says your child might have cystic fibrosis, but we just have to check to be sure, it's probably worse than if I sit down and talk with them, speak with them after I've had a relationship for several years" (F2)
	"I don't know enough to counsel parents and they usually have a lot more questions than I can answer .... I don't know enough about all the statistics and the next steps, and where to go and what bloodwork they have to do. So I find it easier for the person from the newborn screening to call them" (P4)
System design	"Really, like not beyond textbook knowledge and, you know, I've never had that result .... So when I saw [the result], I panicked a little—oh my god—I really don't know anything about this condition, but it didn't matter because I got such good thorough information, I felt like I could convey the basics to my client. And I was able to refer her to the professionals quickly so it was just seamless; it was a really good experience" (M2)
	"So I get a panicked call at about 3 PM or so in the afternoon, from the dad .... 'We got some call about something to do with CF in [child's name].' I had not received any results yet ... I'd received no information, like nothing. They got the call from [NSO] saying they're being referred, so they're in like a total panic .... And so it just, it was a bit of a challenging position for me .... I'm scrambling trying to get information before I call them back, in the middle of clinic. Had I received the result beforehand ... a couple of hours in an evening to figure it out before I called the family to tell them, or before I heard from them" (F4)

CF—cystic fibrosis, F—family physician, M—midwife, NSO—Newborn Screening Ontario, P—pediatrician, PCP—primary care practitioner.

Conclusion

In the context of NBS for CF, PCPs place importance on their involvement in notifying families of screen-positive results. Point-of-care information is essential to facilitating this process. Close links between screening and diagnostic laboratories and PCPs, as well as ongoing educational initiatives in primary care, will be essential for effective delivery of genomic medicine.

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Contributors

All authors were involved in the design of the study and reviewed and edited the manuscript. **Drs Hayeems, Carroll, Miller, Bombard, Potter,** and **Guttmann** and **Ms Barg** were involved in the analysis and **Drs Hayeems, Carroll, Miller, Bombard, Potter,** and **Guttmann** were involved in writing the manuscript.

Competing interests

None declared

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