

Primary care role in expanded newborn screening

After the heel prick test

Robin Z. Hayeems PhD Fiona A. Miller PhD June C. Carroll MD Julian Little PhD Judith Allanson MD
Jessica P. Bytautas Pranesh Chakraborty MD Brenda J. Wilson PhD

Abstract

Objective To examine the role of primary care providers in informing and supporting families who receive positive screening results.

Design Cross-sectional survey.

Setting Ontario.

Participants Family physicians, pediatricians, and midwives involved in newborn care.

Main outcome measures Beliefs, practices, and barriers related to providing information to families who receive positive screening results for their newborns.

Results A total of 819 providers participated (adjusted response rate of 60.9%). Of the respondents, 67.4% to 81.0% agreed that it was their responsibility to provide care to families of newborns who received positive screening results, and 64.2% to 84.8% agreed they should provide brochures or engage in general discussions about the identified conditions. Of the pediatricians, 67.3% endorsed having detailed discussions with families, but only 24.1% of family physicians and 27.6% of midwives endorsed this practice. All provider groups reported less involvement in information provision than they believed they should have. This discrepancy was most evident for family physicians: most stated that they should provide brochures (64.2%) or engage in general discussions (73.5%), but only a minority did so (15.3% and 27.7%, respectively). Family physicians reported insufficient time (42.2%), compensation (52.2%), and training (72.3%) to play this role, and only a minority agreed they were up to date (18.5%) or confident (16.5%) regarding newborn screening.

Conclusion Providers of primary newborn care see an information-provision role for themselves in caring for families who receive positive newborn screening results. Efforts to further define the scope of this role combined with efforts to mitigate existing barriers are warranted.

EDITOR'S KEY POINTS

- Expanded newborn screening has substantially increased the number of positive screening results, prompting attention to the role of primary care providers in informing and supporting families who receive these results. Primary care providers can increasingly expect to be involved in tracking or notifying families with time-sensitive positive screening results, and discussing false-positive and carrier results in the context of routine well-baby care.
- Most providers agreed that it was their responsibility to provide care to families who received positive newborn screening results (67.4% of family physicians, 81.0% of pediatricians, and 71.6% of midwives). Specifically, 70.6% of providers agreed that, once positive screening results were available, they should provide brochures about conditions identified through newborn screening to parents, and 80.0% agreed that they should engage in general discussions with families about these conditions.
- In the context of caring for families of infants who receive positive screening results, this study endorses an information-provision role for primary care providers, efforts to mitigate barriers to pursuing this role, and more fulsome inquiry into defining the actual scope of this role.

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Rôle des intervenants de première ligne dans les nouveaux tests de dépistage pour les nouveau-nés

Au-delà du test néonatal du buvard

Robin Z. Hayeems PhD Fiona A. Miller PhD June C. Carroll MD Julian Little PhD Judith Allanson MD
Jessica P. Bytautas Pranesh Chakraborty MD Brenda J. Wilson PhD

Résumé

Objectif Déterminer le rôle des intervenants de première ligne pour informer et aider les familles qui reçoivent des résultats de dépistage positifs.

Type d'étude Enquête transversale.

Contexte L'Ontario.

Participants Médecins de famille, pédiatres et sages-femmes participant aux soins des nouveau-nés.

Principaux paramètres à l'étude Croyances, façons de faire et obstacles en rapport avec l'information donnée aux familles qui reçoivent des résultats de dépistage positifs pour leur nouveau-né.

Résultats Un total de 819 intervenants ont répondu (taux de réponse ajusté: 60,9%). Parmi les répondants, de 67 à 81,0% reconnaissaient qu'il était de leur responsabilité d'accompagner les familles de nouveau-nés ayant eu des résultats positifs au dépistage et de 64,2 à 84,8% étaient d'avis qu'ils devraient fournir des brochures sur les conditions identifiées ou en discuter de façon générale. Parmi les pédiatres, 67,3% déclaraient avoir des discussions détaillées avec les familles, alors que seulement 24,1% des médecins de famille et 27,6% des sages-femmes le faisaient. Tous les groupes d'intervenants disaient donner moins d'information que ce qu'ils croyaient qu'ils auraient dû. Cela était particulièrement évident pour les médecins de famille: la plupart déclaraient qu'ils auraient dû distribuer des brochures (64,2%) ou entreprendre des discussions générales (73,5%), mais seulement une minorité le faisaient (15,3 et 27,6%, respectivement). Les médecins de famille disaient manquer de temps (42,2%), de rémunération (52,2%) et de formation (72,3%) pour jouer ce rôle, et seulement une minorité se considéraient bien informés (18,5%) et confiants (16,5%) en ce qui concerne le dépistage chez le nouveau-né.

Conclusion Les intervenants qui prodiguent des soins primaires aux nouveau-nés croient qu'ils ont un rôle d'information à jouer auprès des familles qui reçoivent des résultats positifs d'un dépistage néonatal. Il serait opportun de mieux définir les limites de ce rôle mais aussi de minimiser les obstacles existants.

POINTS DE REPÈRE DU RÉDACTEUR

- L'addition de nouveaux tests de dépistage pour les nouveau-nés a considérablement augmenté le nombre de résultats positifs, soulignant ainsi le rôle éventuel des soignants de première ligne pour fournir information et soutien aux familles qui reçoivent de tels résultats. Les intervenants de première ligne doivent s'attendre à être de plus en plus chargés de retracer et d'avertir les familles qui ont des résultats de dépistage positifs et de discuter des faux positifs et des porteurs à l'occasion des soins de routine des bébés.

- La plupart des intervenants étaient d'avis qu'il leur appartenait de s'occuper des familles qui reçoivent des résultats positifs du dépistage de leur nouveau-né (67,4% des médecins de famille, 81,0% des pédiatres et 71,6% des sages-femmes). Plus précisément, 70,6% des intervenants pensaient qu'en présence de résultats positifs, ils devraient fournir aux parents des brochures sur la condition révélée par le dépistage, tandis que 80,0% d'entre eux disaient qu'ils devraient en discuter avec les familles.

- À propos des familles qui reçoivent des résultats positifs du dépistage d'un nouveau-né, les auteurs croient que les intervenants de première ligne ont pour rôle d'informer les parents et ils proposent de minimiser les obstacles qui les empêchent, en plus de mieux définir les limites de ce rôle.

Cet article a fait l'objet d'une révision par des pairs.
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Developments in genetic technologies coupled with a growing understanding of the role of genetics in disease mean that primary care providers will play an increasingly important role in the provision of genetic services.¹⁻⁵ Several initiatives are under way to address the well documented gaps in genetics-related training, as well as the at-times limited clinical value and financial disincentives perceived by primary care providers when faced with delivering genetic services.⁶⁻¹⁵

Newborn blood spot screening, commonly known as the *heel prick test*, presents yet another context in which primary care physicians will be increasingly involved in genetic medicine. While these programs have historically screened for a few rare disorders, developments in technology coupled with parent and professional advocacy have led to substantial growth.¹⁶ In Ontario, since 2005, the newborn screening panel has expanded from screening for 2 to screening for 29 disorders, increasing the number of positive screening results from approximately 60 to approximately 1400 per year.¹⁷ In addition to a potential role in informing women about newborn screening during prenatal care,¹⁸ primary care providers will be called on in the postpartum context.

In the United States (US), some work has been done to define these roles. As primary care providers are typically the first to respond to positive test results,¹⁹ and as their responses affect the family's psychosocial experience,²⁰ effective communication and management of results are paramount.²¹ The American Academy of Pediatrics²² has articulated that primary care pediatricians are responsible for ensuring that newborn screening has been conducted; educating parents regarding positive screening results (including positive carrier results) and further diagnostic steps; and coordinating care for children identified as having serious disorders. Empiric data reflecting on the implementation of these roles suggest that providers are committed to assuming them, but require enhanced training as well as communication and procedural tools.^{21,23-26} In Canada, newborn screening programs operate less consistently than they do in the US, and ensuing roles for primary care providers are less well understood. While primary care providers are responsible for the recall of infants with positive screening results and the coordination of confirmatory testing in several provinces (eg, British Columbia, Alberta, Saskatchewan, Manitoba, Nova Scotia)²⁷⁻³¹ akin to the US, in other provinces (eg, Ontario), the newborn screening program plays a central role in recalling infants with positive screening results and coordinating confirmatory testing. By design, primary care providers are directly involved in this process if the family is far from a tertiary care centre (eg, in northern Canada), or if the tertiary care centre contacts the family's provider to coordinate initial notification and recall.³² How this system of care is operating in real time is not well understood.

Given the known implementation barriers that have been reported in the US context^{21,23-26} and the more varied and less well understood role of primary care providers in Canada, this article explores the scope of primary care providers' role in care after newborn screening. While all of the roles delineated by the American Academy of Pediatrics²² warrant investigation, we present a focus on the primary care provider's educational role in the early postnatal period. We contribute preliminary evidence on providers' beliefs and practices specific to information provision, subsequent to families' receipt of positive screening results in newborns.

METHODS

Detailed methods have been published elsewhere.^{18,33,34} We report data from a cross-sectional survey of a stratified random sample of Ontario health care providers about their perceived roles and responsibilities related to care before and after newborn screening. Specifically, this paper reports on the beliefs and practices of family physicians, pediatricians, and midwives in the early postnatal period. While postnatal midwifery care is limited to 6 weeks, we included midwifery data because positive newborn screening results are available and warrant confirmatory action within this time period. Family physicians and pediatricians were identified using MDSelect, a Canadian medical directory made available by the Canadian Medical Association. Midwives were identified using directories from the College of Midwives of Ontario. Assuming a 50% response rate and a 50% frequency for each outcome, a self-administered questionnaire was mailed to a random sample of family physicians (n=729), all pediatricians (n=569), and all midwives (n=339), and was accompanied by a cover letter, a \$2 coffee coupon, and a postage-paid reply envelope. In accordance with the Dillman design,³⁵ up to 5 mailings were completed over an 8-week period. The McMaster Health Sciences Research Ethics Board in Hamilton approved this study.

The questionnaire was developed by a multidisciplinary team based on a literature review, and was pretested with 2 to 3 members of each provider group for validity. The initial 10 to 15 items were provider-group specific, assessing practices and perceived barriers related to newborn screening care. The remaining 69 items were consistent across provider groups. Using categorical responses and 5-point Likert scales measuring strength of agreement or frequency of practice, we assessed knowledge and confidence about newborn screening; practices related to providing information to families about identified conditions; core beliefs about responsibilities related to informing and caring for families of newborns with positive screening results; and demographic characteristics. Data were entered using

Snap Surveys software and analyzed using SPSS, version 18. Descriptive statistics were computed for all variables, including frequency counts and percentages. We collapsed the Likert scales used to assess providers' practices into 3 levels (consistently or usually; sometimes; rarely or never) and those used to assess core beliefs and perceived barriers into 3 levels (strongly agree or agree; neutral; disagree or strongly disagree).

RESULTS

A total of 819 surveys were completed (adjusted response rate of 60.9%; **Table 1**). A further 109 providers (47 family physicians and 62 pediatricians) were excluded from this analysis, as they indicated that they

were not involved in newborn care. Of the remaining 710 respondents, most practised in non-academic centres (83.7%). A minority of midwives and family physicians practised in urban settings (25.1% and 28.6%, respectively), and slightly more than half the pediatricians practised in urban settings (52.1%). Most family physicians and pediatricians used fee-for-service reimbursement (55.4% and 62.6%, respectively), whereas all midwives (100.0%) were compensated for the course of care. Slightly more than half of family physicians were female (56.9%), slightly more than half of pediatricians were male (55.0%), and all midwives were female (100.0%). Most family physicians and pediatricians had been in practice for 10 years or more (61.4% and 65.4%, respectively), whereas most midwives had been in practice for less than 10 years (76.4%) (**Table 2**).

Table 1. Response rates

RESPONDENTS	TOTAL, N	INELIGIBLE,* N	ADJUSTED N [†]	NO. OF COMPLETED SURVEYS		RESPONSE RATE, % (COMPLETED/ADJUSTED)
				RESPONDENTS INVOLVED IN NEWBORN CARE	RESPONDENTS NOT INVOLVED IN NEWBORN CARE [‡]	
Family physicians	729	144	585	249	47	50.6
Pediatricians	569	136	433	211	62	63.0
Midwives	339	13	326	250	0	76.7
Total	1637	293	1344	710	109	60.9

*Could not be located.

[†]Total sample minus ineligible sample.

[‡]Included in response rate calculations but excluded from analyses.

Table 2. Summary of respondent characteristics: All characteristics are self-defined.

CHARACTERISTIC	FAMILY PHYSICIANS (N = 249), N (%)	PEDIATRICIANS (N = 211), N (%)	MIDWIVES (N = 250), N (%)	TOTAL (N = 710), N (%)
Practice setting				
• Academic	24* (9.8)	64 [†] (30.6)	26 [‡] (10.5)	114 (16.3)
• Non-academic	220 (90.2)	145 (69.4)	221 (89.5)	586 (83.7)
Method of reimbursement				
• Fee-for-service	138 (55.4)	132 (62.6)	0 (0.0)	270 (38.0)
• Non-fee-for-service	111 (44.6)	79 (37.4)	250 (100.0)	440 (62.0)
Practice location				
• Urban	70 [§] (28.6)	110 (52.1)	62 [‡] (25.1)	242 (34.4)
• Non-urban	175 (71.4)	101 (47.9)	185 (74.9)	461 (65.6)
Sex				
• Male	106 [†] (43.1)	116 (55.0)	0 (0.0)	222 (31.4)
• Female	140 (56.9)	95 (45.0)	250 (100.0)	485 (68.6)
Years in practice				
• ≥ 10	153 (61.4)	138 (65.4)	59 (23.6)	350 (49.3)
• < 10	96 (38.6)	73 (34.6)	191 (76.4)	360 (50.7)

*Information missing for 5 respondents.

[†]Information missing for 2 respondents.

[‡]Information missing for 3 respondents.

[§]Information missing for 4 respondents.

Most providers agreed that it was their responsibility to provide care to families who received positive newborn screening results (67.4% of family physicians, 81.0% of pediatricians, and 71.6% of midwives). Specifically, 70.6% of providers agreed that, once positive screening results were available, they should provide brochures about conditions identified through newborn screening to parents, and 80.0% agreed that they should engage in general discussions with families about these conditions. While most pediatricians (67.3%) endorsed having detailed discussions with families about identified conditions, only a minority of family physicians and midwives (24.1% and 27.6%, respectively) endorsed this practice. All provider groups reported engaging less in these information-provision practices than they agreed that they should. This discrepancy was most apparent for family physicians. Whereas most family physicians agreed that they should provide brochures (64.2%) or have general discussions (73.5%) with families about conditions identified through newborn screening, only a minority reported doing so (15.3% and 27.7%, respectively) (Table 3).

Family physicians faced barriers across all measured domains. Most family physicians agreed that they had insufficient time (42.2%), compensation (52.2%), and training (72.3%) to communicate with families of newborns with positive screening results, and only a minority agreed that they were up to date regarding newborn screening (18.5%) or confident to explain it to parents (16.5%). More than a third of pediatricians and midwives reported that they had insufficient compensation, while 18.4% of midwives and 32.2% of pediatricians reported facing time barriers. A substantially lower proportion of pediatricians (33.6%) reported training barriers to providing care for infants with positive screening results, and most agreed that they were up to date (57.8%) or confident (62.6%) with respect to newborn screening (Table 4).

DISCUSSION

Expanded newborn screening increases both the opportunity and the need for the involvement of primary care providers. Specifically, there is an opportunity to enhance primary care providers' educational role with infants who have positive screening results, through notifying the patients' families of initial positive screening results, or through care for the large proportion of infants with positive screening results who prove to have false-positive results or to be carriers upon confirmatory testing.⁵ Yet newborn screening policy and service attention has focused on the coordination of laboratory and specialist capacity. In 2006, the American Academy of Family Physicians voiced concern that a key national report¹⁶ on expanded newborn screening "shows relatively little

recognition of the role of primary care clinicians, who care for newborns and their mothers and to whom families will turn to sort out positive test results."³⁶

Despite limited policy attention, our data suggest that most primary care providers see an educational role for themselves in caring for the growing number of families who receive positive screening results. As might be expected, family physicians and midwives appeared committed to a general educational role for these families, while pediatricians perceived a responsibility for assuming a more specialized educational role. However, family physicians perceived more constraints than the other 2 provider groups in assuming this role because of training and structural barriers. This is particularly concerning given family physicians' central role in well-baby care, especially outside urban areas, where they are effectively the only providers of such care.³⁷

This study provides important insights regarding the primary care provider's role in newborn screening care, and genetic medicine more generally. First, while some point to persistent scepticism among primary care providers regarding the importance of genetics,^{2,38-40} our study suggests that newborn screening-related primary care might be different. Perhaps there is a strong sense of professional responsibility because it so clearly concerns a fundamental primary care role—that of well-baby care. Or perhaps this is because genetic information, in the context of newborn screening, can have a demonstrably positive effect on clinical outcomes. Second, our study points to the importance of challenges beyond those that are training-related that must be overcome if primary care providers are to support families of infants who receive positive screening results. In addition to educational resources to build core competencies,^{5-11,41-44} primary care providers might require more structural resources, involving time and compensation.

Limitations

The survey instrument was designed to explore several prenatal and postnatal components of newborn screening care and was launched soon after the newborn screening program in Ontario expanded. Thus, we were constrained in the detail we could capture related to the extent to which newborn screening had affected respondents' clinical case mix or volume. Further, with respect to beliefs related to perceived responsibility to care for families with infants who received positive screening results, we did not specify the type of positive screening result, limiting our ability to identify which positive screening context (initial positive, true positive, false positive) respondents were reflecting on when answering the questionnaire items. With respect to the relative frequency with which they provided information, some respondents might have reported how often they were presented with the opportunity to do this rather than the frequency with which they made use

Table 3. Respondents' core beliefs and practices

QUESTION	FAMILY PHYSICIANS (N = 249), N (%)	PEDIATRICIANS (N = 211), N (%)	MIDWIVES (N = 250), N (%)	TOTAL (N = 710), N (%)
Core beliefs				
Before screening results are available				
• Feel responsible for providing care to families who receive positive newborn screening results				
- Agree	168 (67.4)	171 (81.0)	179 (71.6)	518 (73.0)
- Neutral	43 (17.3)	9 (4.3)	20 (8.0)	72 (10.1)
- Disagree	17 (6.8)	5 (2.4)	35 (14.0)	57 (8.0)
- Missing	21 (8.4)	26 (12.3)	16 (6.4)	63 (8.9)
Once positive screening results are available				
• Should provide brochures to parents about a condition identified through newborn screening				
- Agree	160 (64.2)	154 (73.0)	187 (74.8)	501 (70.6)
- Neutral	33 (13.2)	31 (14.7)	21 (8.4)	85 (12.0)
- Disagree	48 (19.3)	21 (10.0)	34 (13.6)	103 (14.5)
- Missing	8 (3.2)	3 (1.4)	8 (3.2)	19 (2.7)
• Should have a general discussion with parents about a condition identified through newborn screening				
- Agree	183 (73.5)	179 (84.8)	206 (82.4)	568 (80.0)
- Neutral	23 (9.2)	17 (8.1)	12 (4.8)	52 (7.3)
- Disagree	35 (14.0)	12 (5.7)	23 (9.2)	70 (9.8)
- Missing	8 (3.2)	3 (1.4)	9 (3.6)	20 (2.8)
• Should have a detailed discussion with parents about a condition identified through newborn screening				
- Agree	60 (24.1)	142 (67.3)	69 (27.6)	271 (38.2)
- Neutral	45 (18.1)	33 (15.6)	23 (9.2)	101 (14.2)
- Disagree	137 (55.0)	32 (15.2)	150 (60.0)	319 (44.9)
- Missing	7 (2.8)	4 (1.9)	8 (3.2)	19 (2.7)
Actual practices				
Once positive screening results are available				
• Provide brochures to parents about a condition identified through newborn screening				
- Consistently or usually	38 (15.3)	55 (26.1)	82 (32.8)	175 (24.6)
- Sometimes	20 (8.0)	47 (22.3)	17 (6.8)	84 (11.8)
- Rarely or never	175 (70.3)	105 (49.8)	141 (56.4)	421 (59.8)
- Missing	16 (6.4)	4 (1.9)	10 (4.0)	30 (4.2)
• Have a general discussion with parents about a condition identified through newborn screening				
- Consistently or usually	69 (27.7)	117 (55.4)	116 (46.4)	302 (42.5)
- Sometimes	25 (10.0)	36 (17.1)	10 (4.0)	71 (10.0)
- Rarely or never	137 (55.0)	55 (26.1)	115 (46.0)	307 (43.2)
- Missing	16 (6.4)	3 (1.4)	9 (3.6)	28 (3.9)
• Have a detailed discussion with parents about a condition identified through newborn screening				
- Consistently or usually	39 (15.7)	108 (51.2)	60 (24.0)	207 (29.2)
- Sometimes	28 (11.2)	36 (17.1)	25 (10.0)	89 (12.5)
- Rarely or never	165 (66.3)	64 (30.3)	156 (62.4)	385 (54.2)
- Missing	17 (6.8)	3 (1.4)	9 (3.6)	29 (4.1)

Table 4. Respondents' perceived barriers

BARRIERS	FAMILY PHYSICIANS (N = 249), N (%)	PEDIATRICIANS (N = 211), N (%)	MIDWIVES (N = 250), N (%)	TOTAL (N = 710), N (%)
Insufficient time to care for families who receive positive newborn screening results				
• Agree	105 (42.2)	68 (32.2)	46 (18.4)	219 (30.8)
• Neutral	51 (20.5)	26 (12.3)	48 (19.2)	125 (17.6)
• Disagree	77 (30.9)	95 (45.0)	146 (58.4)	318 (44.8)
• Missing	16 (6.4)	22 (10.4)	10 (4.0)	48 (6.8)
Insufficient compensation to care for families who receive positive newborn screening results				
• Agree	130 (52.2)	77 (36.5)	85 (34.0)	292 (41.1)
• Neutral	69 (27.7)	53 (25.1)	69 (27.6)	191 (26.9)
• Disagree	33 (13.2)	57 (27.0)	85 (34.0)	175 (24.6)
• Missing	17 (6.8)	24 (11.4)	11 (4.4)	52 (7.3)
Insufficient training to care for families who receive positive newborn screening results				
• Agree	180 (72.3)	71 (33.6)	165 (66.0)	416 (58.6)
• Neutral	33 (13.2)	27 (12.8)	25 (10.0)	85 (12.0)
• Disagree	21 (8.4)	89 (42.2)	50 (20.0)	160 (22.5)
• Missing	15 (6.0)	24 (11.4)	10 (4.0)	49 (6.9)
Up to date on conditions included in Ontario's newborn screening program				
• Agree	46 (18.5)	122 (57.8)	122 (48.8)	290 (40.8)
• Neutral	58 (23.3)	34 (16.1)	35 (14.0)	127 (17.9)
• Disagree	138 (55.4)	43 (20.4)	91 (36.4)	272 (38.3)
• Missing	7 (2.8)	12 (5.7)	2 (0.8)	21 (3.0)
Confident in ability to explain newborn screening to parents				
• Agree	41 (16.5)	132 (62.6)	153 (61.2)	326 (45.9)
• Neutral	59 (23.7)	35 (16.6)	46 (18.4)	140 (19.7)
• Disagree	142 (57.0)	32 (15.2)	49 (19.6)	223 (31.4)
• Missing	7 (2.8)	12 (5.7)	2 (0.8)	21 (3.0)

of such opportunities (as was intended). Further, we are unable to infer respondents' actual interpretation of "caring for families" or the content they perceived to be contained within a general discussion or a detailed discussion. Lack of clarity on these definitions might have contributed to response-item overlap; however, our emphasis on provider group differences over absolute proportions remains instructive.⁴⁵ Now that this exploratory study has highlighted primary care providers' general commitment to this expanding domain of care, we hope that future studies will correct these limitations and explore these issues in greater depth.

Conclusion

In the context of caring for families of infants who receive positive screening results, this study endorses an information-provision role for primary care providers, efforts to mitigate barriers to pursuing this role, and more fulsome inquiry into defining the actual scope of this role.

Dr Hayeems is Postdoctoral Fellow and **Dr Miller** is Associate Professor in the Institute of Health Policy, Management and Evaluation at the University of Toronto in Ontario. **Dr Carroll** is Associate Professor in the Department of Family and Community Medicine at the University of Toronto and a family physician in the Granovsky Gluskin Family Medicine Centre at Mount Sinai Hospital in Toronto. **Dr Little** is Associate Professor and Chair of the Department of Epidemiology and Community Medicine at the University of Ottawa in Ontario. **Dr Allanson** is Chief of the Department of Genetics at the Children's Hospital of Eastern Ontario in Ottawa and the Department of Pediatrics at the University of Ottawa. At the time of this study, **Ms Bytautas** was Research Officer in the Institute of Health Policy, Management and Evaluation at the University of Toronto. **Dr Chakraborty** is Clinical Metabolic Geneticist in the Department of Genetics and Director of Newborn Screening Ontario at the Children's Hospital of Eastern Ontario. **Dr Wilson** is Professor in the Department of Epidemiology and Community Medicine at the University of Ottawa.

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Contributors

Drs Miller and Hayeems conceived the study and led the analysis and writing. **Drs Carroll and Little** contributed to the conceptual plan for the study and were core members of the analytic and writing teams. **Dr Allanson, Ms Bytautas, Dr Chakraborty, and Dr Wilson** reviewed the conceptual plan for the study and supported its analysis and writing.

Competing interests

None declared

Correspondence

Dr Fiona A. Miller, Department of Health Policy, Management and Evaluation, University of Toronto, 155 College St, 4th Floor, Toronto, ON M5T 3M6; telephone 416 978-3703; fax 416 978-7350; e-mail fiona.miller@utoronto.ca

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