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A secondary benefit: the reproductive impact of carrier results from newborn screening for cystic fibrosis

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Abstract

Purpose—Newborn screening (NBS) for cystic fibrosis (CF) can identify carriers, which is considered a benefit that enables reproductive planning. We examined the reproductive impact of carrier result disclosure from NBS for CF.

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Methods—We surveyed mothers of carrier infants after NBS (Time-1) and one-year later (Time-2) to ascertain intended and reported communication of their infants' carrier results to relatives, carrier testing for themselves/other children and reproductive decisions. A sub-sample of mothers was also interviewed at Time-1 and Time-2.

Results—Response rate was 54%. Just over half (55%) of mothers carrier tested at Time-1; a further 40% of those who intended to test at Time-1 tested at Time-2. Carrier result communication to relatives was high (92%), but a majority of participants did not expect the results to influence family planning (65%). All interviewed mothers valued learning their infants' carrier results. Some had carrier testing and shared results with family. Others did not use the results or used them in unintended ways.

Conclusion—While mothers valued learning carrier results from NBS, they reported moderate uptake of carrier testing and limited influence on family planning. Our study highlights the secondary nature of the benefit from disclosing carrier results from NBS.

Keywords

newborn screening; cystic fibrosis; false positive; carrier status; carrier testing

INTRODUCTION

Newborn screening (NBS) aims to reduce childhood morbidity and mortality through early identification and treatment of affected infants.[1] This is considered the primary benefit of NBS programs, encouraging universal screening of infants for many disorders worldwide. Secondary benefits may also accrue through NBS such as benefits to the family and society, where NBS can inform family planning ('reproductive benefit') or advance the understanding of disease. Traditionally, these secondary benefits have not been sufficient to justify NBS.[2] However, recent scholarly discourse has highlighted reproductive benefit as an increasingly prominent goal of NBS, particularly when the clinical goal of identifying a treatable condition may not be assured.[3–6] One way reproductive benefits arise through NBS is through the generation of carrier results.

NBS for cystic fibrosis (CF) using typical testing protocols identifies many unaffected carriers of one CF mutation in addition to affected infants; indeed, the majority of infants with false positive CF NBS results are identified as CF carriers during confirmatory testing.[7] In Ontario, CF NBS involves a two-step process of measuring immunoreactive trypsinogen (IRT) followed by screening the CF transmembrane regulator (CFTR) gene for 39 mutations.[8] Confirmatory sweat chloride testing is then performed on screen positive infants who are subsequently classified as true positive, false positive, or inconclusive (i.e. genetic variants of uncertain significance +/− borderline sweat chloride; Table 1). Both parents of carrier newborns are offered genetic counselling and carrier testing at no charge as part of the disclosure protocol.

There is long-standing debate about incidental carrier status identification through NBS. First, from an ethical perspective, this information is not typically available without informed consent,[9] and carrier testing is not usually pursued in minors.[10–12] Second, there is

inconsistent evidence about the benefits and harms of CF carrier identification through NBS for parents, including the potential to inform family planning and the risk of psychosocial harms.[13] Studies have shown that communicating carrier results to parents of false-positive infants may cause anxiety.[14,15] While this state tends to be short-lived,[15–19] anxiety about their infants' carrier status persists for a minority of parents, and lead to concern about stigma and the physical health of their carrier child.[18,20,21] Third, parents typically state that they want infant carrier information to know reproductive risks,[19,21–24] yet evidence regarding parental use of that information is equivocal. A minority of parents avoided pregnancy after the disclosure of an infant's carrier status through NBS.[22] (Table 2) Uptake of prenatal diagnosis in subsequent pregnancies ranges from 14–66%, with higher termination rates at 69–100%.[25–27] While the majority of parents share their infants' carrier results with relatives,[18,21] evidence of parents' pursuit of their own carrier testing is inconsistent, varying from 30–85% among parents of carrier infants,[18,21,22,24] with reduced uptake among next degree relatives.[28] (Table 2)

Several studies have documented parents' reproductive attitudes and behaviours following CF identification, [21,25,27,29–33] but these findings are not specific to NBS nor are they longitudinal. Thus, there is limited evidence about the nature or extent of the 'reproductive benefits' of sharing carrier results from NBS. We examined the uptake of carrier testing, carrier result communication and impact of carrier results on family planning among parents of a prospective cohort of false-positive CF infants identified through NBS. We also examined factors associated with these outcomes. Based on existing evidence,[21,25,33–35] we hypothesized that infants' carrier status and mothers' parity, education and income levels would be associated with increased uptake and intentions to pursue carrier testing for themselves, share the results with relatives and use the results to inform family planning. We also explored participants' attitudes regarding these behaviors to elucidate the variation in uptake of these behaviors.

MATERIALS AND METHODS

Study design

This study is part of a prospective, longitudinal, mixed-methods cohort study designed to investigate the impact of NBS for CF on families, health care providers and health services in Ontario. We received research ethics board approval from the University of Toronto, the Children's Hospital of Eastern Ontario, and the Hospital for Sick Children.

Sample

We recruited all mothers of infants confirmed to be false positive for CF after NBS follow-up at the Hospital for Sick Children during the 18-month data collection period. We excluded infants known to be deceased or in the NICU; those adopted or involved with child welfare; and based on clinical judgment of inappropriateness (e.g., extreme distress, catastrophic events, significant language barriers).

Data collection

Surveys—We collected structured data using self-administered questionnaires with a modified Dillman approach [36] where up to three contacts were made. The Time-1 survey was sent to mothers with false positive infants 4–8 weeks after confirmatory testing; confirmatory testing typically occurred when infants were approximately 4 weeks old. The Time-2 survey occurred one year later. Completion and return of the questionnaire package constituted consent to participate.

Questionnaire design—The questionnaire was developed by a multidisciplinary team based on literature review.[18,20–22] We pre-tested the questionnaire with new parents, recruited from the Greater Toronto Area (N=11) through an online mothers' group to assess comprehension, face and content validity.

Interviews—We conducted semi-structured, open-ended interviews with the subsample of mothers of false positive infants from the Time-1 and Time-2 cohorts who indicated their willingness on questionnaires and provided informed consent for in-person or telephone interviews. Interviews explored experiences regarding: uptake of carrier testing; sharing carrier results with relatives and with their carrier infant; and family planning.

Measures

The measures specific to reproduction included: (1) carrier status of the infant, (2) uptake of carrier testing by mothers and their partners, (3) communication of infants' carrier results to relatives, (4) influence of carrier results on family planning; and, (5) carrier testing of other children. Each measure in the questionnaire is detailed in Appendix 1.

Analysis

Surveys—We calculated the proportion of respondents who indicated yes/no to the measures above. We used Chi-square and Fisher Exact tests to test hypotheses of associations between participant characteristics and these measures. We considered two-sided *p*-values of 0.05 or less to indicate statistical significance. Data were managed and analyzed using SPSS 16.0.0 (SPSS Inc., Chicago, IL, USA).

We report cross-sectional analyses for Time-1 and longitudinal results for the subsample of respondents who completed both Time-1 and Time-2 questionnaires. Time-1 questionnaires compare across several study groups (carrier, non-carrier, other/uncertain); Time-2 results are restricted to mothers of carriers as skip patterns prompted non-carrier/other mothers to skip sections pertaining to reproductive risk.

Interviews—Interviews were taped, transcribed and coded. We used a thematic approach, applying and modifying pre-existing codes from the interview guide pertaining to carrier testing, family communication and family planning, and allowed new themes to emerge from the data using constant comparison [37]. We used Time-1 interviews to identify themes and then searched for confirming/disconfirming evidence in Time-2 interviews. No new themes arose in Time-2 interviews and therefore these data are not shown.

RESULTS

Response rate

We received completed questionnaires from 134 of 246 mothers (54%) at Time-1 and 95 of 216 (44%) at Time-2 (30 fewer mothers were approached at T2 because they declined participation at T1 and were not re-approached at T2 or their survey was returned undelivered). We report data on a total of 131 mothers (Time-1) of whom 74 (Time-2) completed Time-1 and Time-2 surveys and responded to the carrier status question at both T1 and T2.

Carrier status

At Time-1, 77 of 131 mothers (59%) reported that one mutation had been identified in their infant ("mothers of carriers"), 30 (23%) reported no classic mutations ("mothers of non-carriers"), 20 (15%) were "unsure", and 4 (3%) indicated "Other" (i.e., genetic testing was pending). At Time-2, mothers of carriers comprised 77% (57/74) of the sample, with another 18% (13/74) mothers of non-carriers and 5% (4/74) of mothers who were "unsure/don't know" their infant's carrier status (Table 3).

Participants' characteristics

The characteristics of our survey samples are reported in Table 3 and appear similar to those of the CF carrier population reported in other studies except that our samples had higher education levels [18,21,22]. There were no significant differences in characteristics between mothers of carriers and other mothers at Time-1 and Time-2, except that at Time-1, mothers of carriers had higher incomes compared to other mothers.

We interviewed 22 mothers of carriers at Time-1 and 25 at Time-2 (7 were interviewed twice). The majority of Time-1 participants were over 30 years old (18/22; 82%), lived in larger cities (16/22; 73%) and earned over $80,000 (18/21; 86%).

Survey results

Carrier Testing Uptake—At Time-1, carrier testing uptake was reported by 55% (42/77) of mothers of carriers (Table 4). Four of 30 (13%) mothers of non-carriers and 10/24 (42%) mothers in the "other/don't know" category also reported they had been tested. Mothers also reported fathers' testing uptake; carrier testing uptake and intentions reported for fathers were similar to mothers' reported uptake and intentions (data not shown).

Follow through on intentions to have carrier testing: Fifteen mothers of carriers who had not had carrier testing at Time-1 intended to and, of those, 6 (40%) were tested by Time-2 (Figure 1). Of the remaining 9, 7 still indicated an intention to test at Time-2 and 2 were unsure. The 11 Time-1 mothers of carriers who did not plan to test or were unsure remained such at Time-2.

Influence of carrier results on family planning—At Time-1, expectations that an infant's carrier result would influence family planning did not differ significantly across groups (p=0.488) (35%, 26/77 mothers of carriers; 27%, 7/30 non-carriers; 25%, 6/24

unsure/other mothers) (Table 4); of these, 18 (69%) mothers of carriers indicated that the results would influence them to have subsequent children, as did 2 (29%) non-carriers and 2 (33%) who were unsure/other (Table 4).

Follow through on expectations for results to influence family planning: Seventeen mothers expected that their child's carrier status would influence family planning at Time-1; 11 expected to have subsequent children, of whom 6 (55%) had or still planned to have more children at Time-2 (Figure 1). The majority (>65%) of mothers of carriers did not expect, or were unsure whether to expect, the results to influence family planning, which remained consistent at follow-up.

Communicating carrier results to relatives—At Time-1, most mothers of carriers had told relatives they may also be carriers (70/77; 92%), as had 32% (9/30) of mothers of non-carriers and 75% (18/24) of those who were "unsure/other" (Table 4).

Follow through on intentions to communicate results to family: Only a minority (3/57; 5%) of mothers of carriers did not tell relatives they may be carriers at Time-1 and, of those, all 3 told relatives at follow up (Figure 1).

Carrier testing among other children—Few mothers had their other children carrier tested at Time-1: 8% (3/37) of mothers of carriers, 6% (1/16) of non-carriers and none of the unsure/other mothers (Table 4).

Factors associated with reproductive behaviours—At Time-1, mothers of carriers were significantly more likely to have carrier testing themselves, express an intention to test and notify relatives of their infant's carrier results compared to other mothers ($p<0.001$) (Table 4). Primipara mothers were more likely than other mothers to have carrier testing and to expect that the results would influence their family planning ($p<0.01$– Table 4 & Supplemental Table 1).

Qualitative results

Our qualitative analysis extends our survey data, suggesting the existence of two groups of mothers. While all mothers identified value in learning their infants' carrier information, some (1) used it in a targeted fashion to inform carrier testing and family communication; others (2) either did not use it or used it in unintended ways.

Reproductive benefit of carrier information—For some mothers, their infants' carrier results were influential in informing family planning for themselves, which motivated parents to pursue their own carrier testing: "*if we were both carriers, we actually kinda decided we wouldn't have another kid.*" (ID#253) Carrier results were also perceived as important for relatives' family planning, which was another motivation to pursue their own carrier testing: *"Once we figured out who was the carrier then we discussed [results] with that side of the family… knowledge is power in this case".* (ID#50) Mothers reported informing first-, second- and third-degree relatives, particularly those planning to have children. Many described the nature of the communication as "*talking to [relatives] about it*", while others forwarded letters provided by the clinics to the family members who were

at risk: "*I have plans to send an email around with the information I got from the hospital.*" (ID#281).

These mothers also valued learning their infants' carrier results for their children's future reproductive planning and partner selection. In most cases, parents planned to discuss the results with their future-adult child, and did not perceive this to be "*too big of an issue*" (ID#198). However, some were concerned that their child would misunderstand the implications or worry: *"I would just want to make sure that she's at a mature enough level that alarm bells don't go off in her head and she starts stressing about, is this a disease I'm going to get".* (ID#141) Learning about infants' carrier status sometimes represented an opportunity to learn whether other children were also carriers, which motivated some mothers to have carrier testing; however, few also tested their other children.

Lack of reproductive benefit of carrier information—Other mothers appreciated receiving their infants' carrier results but did not use the information or use it in an unintended manner. In some instances, this was because they did not plan to have more children. In other instances, the reproductive value of the information was not a focus. This was evident when parents indicated that they pursued carrier testing for themselves out of curiosity or convenience: "*I was just curious"* (ID#282). Mothers in this group also shared their infants' carrier results with both sides of the family without attending to which parent was the carrier, and thus which side of the family was specifically at risk: *"We're letting everybody know, but we didn't feel like we needed to do the testing ourselves to narrow down exactly who to give the information to".* (ID#70) These mothers were also uncertain about sharing the infant's carrier results with their future adult-child or gave it little consideration. Finally, these mothers did not expect the results to inform reproductive planning, noting that they would: "*continue on just like we have"* (ID#182).

DISCUSSION

Our study provides the first prospective, longitudinal mixed-methods data on the reproductive impact of carrier results from NBS for CF. Our results suggest that the reproductive benefits of CF carrier disclosure through NBS among mothers of CF carrier infants are not uniform or consistent. Carrier result communication to relatives was high (92%) and some found the information influential in informing their family planning. But there was moderate carrier testing uptake (55%), and a majority of participants did not expect the results to influence family planning (65%). Interviews also identified a lack of utility in family planning for some because of life stage. Finally, carrier results were sometimes used in unintended ways: some parents tested their other children, and non-carriers informed their relatives that they may be carriers of CF. These actions may prompt unnecessary use of health care services or lead to concern among relatives, in addition to challenging international guidelines on carrier testing of minors. Given the moderate levels of utility and unintended consequences, our study highlights the secondary nature of the benefit arising from the generation of carrier results from NBS.

Our results are consistent with literature reporting that a majority of parents share their infants' carrier results with relatives,[18,21] and that a minority indicate that the results

influence decisions to have more children. [21,22,25,26] Our maternal carrier testing uptake rate reflects the median reported among parents of carrier infants (30–85%; Table 2); this was associated with mothers' parity and their infants' carrier status. Only one other study compared hypothetical and reported reproductive behaviors but these were among parents of children with CF and were restricted to use of prenatal testing and termination of pregnancy. [26] Thus, our study provides novel results on the intended versus actual uptake of carrier testing, results communication and influence on family planning among parents of carrier infants identified through NBS, the associated factors, and provides qualitative insights into the variation in behaviours reported in the literature (summarized in Table 2).

Our study also revealed some unintended consequences of CF carrier results disclosure. First, some mothers "*told everyone*" in their families that they may be CF carriers without confirming which side was at risk. This may create more carrier testing and counseling services than would otherwise be necessary. Second, there appears to be evidence of some confusion since mothers of non-carrier infants also reported pursuing carrier testing (13%) and telling relatives they may be carriers (32%), which is inconsistent with guidelines[38]. These results indicate a need for improved parental understanding of the implications of non-carrier results. Equipping primary care providers with detailed information about NBS, carrier results and their reproductive implications is an additional avenue to ensure patient understanding, as primary care providers often support patients with positive NBS results. Third, a minority of providers appears to be testing other children for carrier status. This reveals fundamental challenges of carrier identification through NBS, which may create disparities in access to carrier information between newborns and their older siblings [39] and conflicts with existing policies and norms that advise against carrier testing of asymptomatic children.[9]

Our results provide timely contributions to the evidence-base about the reproductive impact of sharing carrier results identified through NBS in light of on-going expansions of NBS worldwide. Our findings raise broader questions about the potential reproductive benefit derived from carrier disclosure as a result of NBS, and warrant consideration in policy decisions supporting expanded screening programs. While mothers valued learning their infant's carrier results, our study demonstrates moderate intended and reported testing uptake and limited influence on family planning. These results suggest that carrier results should be considered a secondary benefit, supporting previous calls to consider them "additional" or "limited secondary" benefits.[21,25]

There are several limitations to our study. One year may not be a sufficient time horizon to assess maternal carrier-testing uptake. We did not assess mothers' understanding of carrier status. However, virtually all parents with false-positive results in Ontario are offered free genetic counselling and carrier testing. Thus access to genetic testing and counselling or a lack of understanding of the reproductive implications should not represent major confounders in our study. Further, the interviews provide added support of mothers' understanding of the implications of their infants' carrier status. Finally, while our response rate was modest, it is consistent with, if not higher than, similar population-based surveys among CF cohorts (e.g., 37%[22], 45%[18], 53%[40]). Further, our study provides the first

prospective, longitudinal mixed-methods results on the reproductive impact of carrier results from newborn screening for CF.

Limitations notwithstanding, our study provides a timely contribution to the evidence-base for the 'reproductive benefits' of sharing carrier results to inform NBS policy. Carrier identification through NBS may motivate carrier testing and family communication, but does not necessarily influence family planning or reproductive behaviours. If the influence of carrier status on family planning is a proxy for the value of carrier information obtained through NBS, then this benefit is achieved for only a minority of individuals, which suggests that should it maintain its historic status as a secondary benefit of NBS.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

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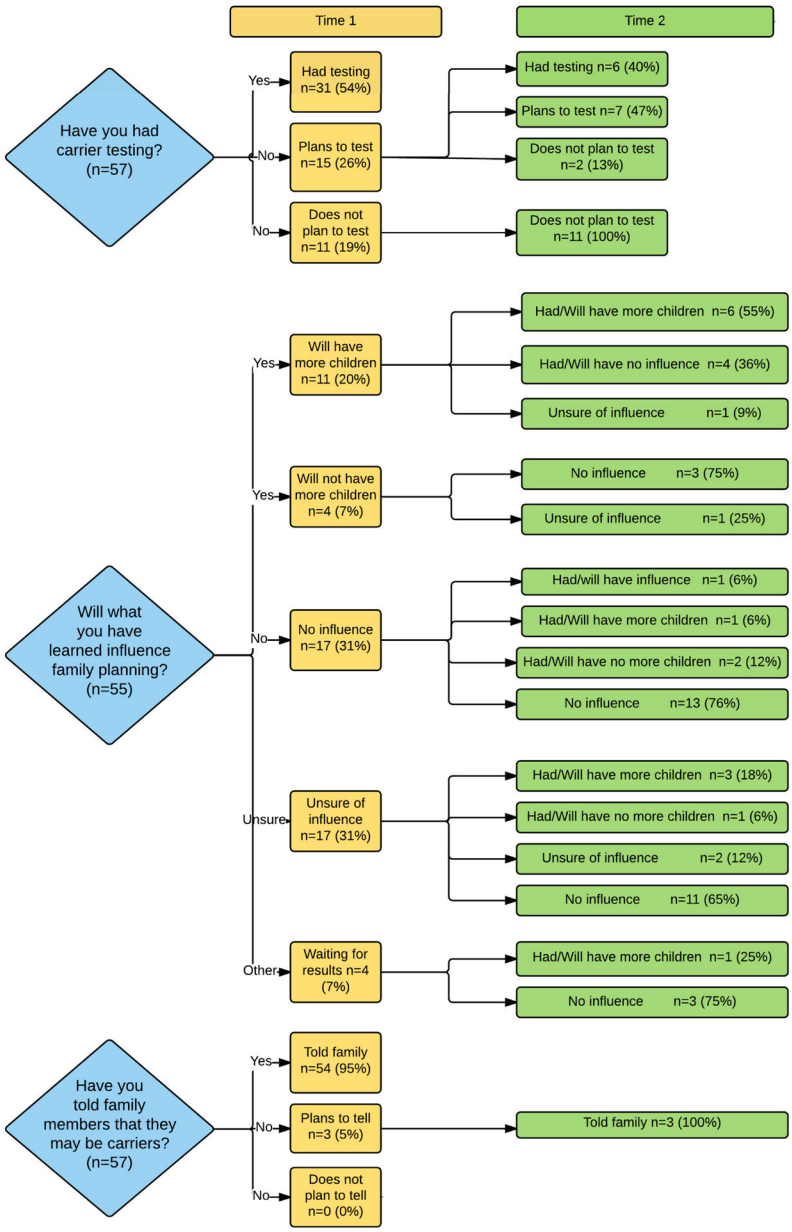

FIGURE 1.
FOLLOW THROUGH WITH INTENTIONS – LONGITUDINAL SURVEY RESULTS

TABLE 1

NEWBORN SCREENING PROTOCOL FOR CF IN ONTARIO

	Screening and diagnostic test results
Inconclusive result	1 CFTR mutation OR IRT > 99.9th centile PLUS sweat chloride 30–59 mmol/L -OR- normal sweat chloride concentration (<30 mmol/L) + ≥1 CFTR mutations of uncertain significance
False positive result	1 CF causing mutation OR IRT > 99th centile PLUS sweat chloride concentration <30 mmol/L
True positive result	2 CF causing mutations OR sweat chloride ≥ 60mmol/L.
Screen negative controls	screen negative for all NBS conditions assessed

* confirmatory testing performed when IRT>96th centile and ≥1 CFTR mutation detected OR IRT>99.9th centile with no CFTR mutations detected

TABLE 2

PREVIOUS EVIDENCE OF REPRODUCTIVE IMPACT OF CF CARRIER IDENTIFICATION

Reference	Country	Would avoid pregnancy	Have avoided pregnancy	Would use PND	Have used PND	Would terminate affected pregnancy	Have terminated affected pregnancy	Shared results with relatives	Had carrier testing
CF diagnosed clinically									
Evers-Kiebooms et al 1990 (34)	Belgium	54%	63%[1]	66%	53%	45%	50%	-	-
Watson et al 1992 (33)	U.K.	64%	-	74%	-	44%	-	-	-
Wertz et al 1992 (32)	U.S.A.	56%	62%	77%[2]	77%[3]	28%	-	-	-
Henneman et al 2001 (40)	The Netherlands	57%[4]	32%	76%	72%	57%	29%	-	-
CF diagnosed through NBS									
Mischler et al 1998 (21)	U.S.A.	-	52%[5]	-	26%[6]	-	0%[7]	-	-
Dudding et al 2000 (25)	Australia	-	59%	-	66%	69%[8]	69%[9]	-	-
Scotet et al 2000 (27)	France	-	-	-	34%	-	100%	-	-
Sawyer et al 2006 (26)	Australia	48%	39%	82%	55%	56%	100%[10]	-	-
Carrier identified through NBS									
Mischler et al 1998 (21)	**U.S.A.**	**4–17%**[11]	-	-	-	-	-	**88%**	**58%**[12]
Ciske et al 2001 (22)	**U.S.A.**	-	**36%**	**9%**	**14%**	-	-	-	**30%**[13]
Wheeler et al 2001 (24)	**U.S.A.**	-	-	-	-	-	-	-	**75%**[14]
Lagoe et al 2005 (35)	**U.S.A.**	-	-	-	-	-	-	-	**38%**
Lewis et al 2006 (18)	**Australia**	**12–13%**[15]	-	-	-	-	-	**71%**[16]	**53**[17]**–85%**[18]

[1] Average across birth rank of the CF-affected child

[2] Study reports combined results for would use & have used PND

[3] Study reports combined results for would use & have used PND

[4] Parents indicated CF diagnosis would 'influence subsequent family planning'

[5] 74% of these already had one child

[6] In first subsequent pregnancy

[7] Of 3 affected pregnancies identified; all were carried to term

[8] Study reports combined results for would & have terminated

[9] Study reports combined results for would & have terminated

[10] Of 5 affected pregnancies identified; all were terminated

[11] Depending on whether IRT or IRT/DNA testing algorithms used

[12] Where both parents were tested

[13] Where both parents were tested – higher when one partner tested first: 42% of mothers & 34% of fathers

[14] Where both parents were tested

[15] Depending on cohort surveyed (1996/7 or 2001)

[16] In earlier (1996/7) cohort surveyed

[17] In earlier (1996/7) cohort surveyed

[18] In later (2001) cohort surveyed

TABLE 3

DEMOGRAPHIC CHARACTERISTICS OF SURVEY PARTICIPANTS

	Survey participants									
	Time 1					Time 2				
	Total (*n*=131)	Mothers reporting 1 mutation (*n*=77)	Mothers reporting no mutations (*n*=30)	Mothers who don't know/ other (*n*=24)	*p*-value	Total (*n*=74)	Mothers of carriers (*n*=57)	Mothers of non-carriers (*n*=13)	Don't know carrier status (*n*=4)	*p*-value
Age										
25 and under	8 (6%)	5 (7%)	1 (3%)	2 (8%)	.07[a]	2 (3%)	2 (3%)	0 (0%)	0 (0%)	.20[a]
26–30	28 (21%)	17 (22%)	2 (7%)	9 (38%)		12 (16%)	11 (19%)	0 (0%)	1 (25%)	
31–35	59 (45%)	34 (44%)	19 (63%)	6 (25%)		30 (41%)	22 (39%)	8 (62%)	0 (0%)	
36+	36 (28%)	21 (27%)	8 (27%)	7 (29%)		30 (41%)	22 (39%)	5 (39%)	3 (75%)	
City size										
100,000 +	92 (71%)	50 (66%)	25 (83%)	17 (71%)	.20[b]	50 (69%)	37 (66%)	9 (69%)	4 (100%)	.52[a]
< 100,000	38 (29%)	26 (34%)	5 (17%)	7 (29%)		23 (32%)	19 (34%)	4 (31%)	0 (0%)	
First child										
Yes	70 (53%)	41 (53%)	14 (47%)	15 (63%)	.51[b]	40 (54%)	32 (56%)	6 (46%)	2 (50%)	.83[a]
No	61 (47%)	36 (47%)	16 (53%)	9 (38%)		34 (46%)	25 (44%)	7 (54%)	2 (50%)	
Marital status*										
Married or common law	121 (92%)	72 (94%)	28 (93%)	21 (88%)	.59[a]	72 (97%)	55 (97%)	13 (100%)	4 (100%)	1.0[a]
Other	10 (8%)	5 (7%)	2 (7%)	3 (13%)		2 (3%)	2 (4%)	0 (0%)	0 (0%)	
Education, highest level completed										
High school or less	18 (14%)	5 (7%)	5 (17%)	8 (33%)	.01[a]	7 (10%)	5 (9%)	1 (8%)	1 (25%)	.20[a]
College or CEGEP	53 (41%)	35 (46%)	10 (33%)	8 (33%)		27 (37%)	24 (42%)	2 (15%)	1 (25%)	
Undergrad	27 (21%)	19 (25%)	8 (27%)	0 (0%)		17 (23%)	10 (18%)	6 (46%)	1 (25%)	
Grad or professional	33 (25%)	18 (23%)	7 (23%)	8 (33%)		23 (31%)	18 (32%)	4 (31%)	1 (25%)	
Income										
Under $80,000	49 (39%)	22 (29%)	13 (45%)	14 (64%)	.01[b]	20 (29%)	14 (26%)	3 (25%)	3 (75%)	.12[a]
$80,000 +	77 (61%)	53 (71%)	16 (55%)	8 (36%)		50 (71%)	40 (74%)	9 (75%)	1 (25%)	

[a] Fisher's exact *p*-value

[b] Chi-square *p*-value

TABLE 4

CARRIER TESTING, FAMILY COMMUNICATION & FAMILY PLANNING - TIME 1 CROSS-SECTIONAL RESULTS

	Carrier status of infant						
	Carrier (n=77)[a]		Non-Carrier (n=30)[a]		Other* (n=24)[a]		p-value
	n	%	n	%	n	%	
Carrier testing							
Have you had carrier testing?							
Yes	42	55%	4	13%	10	42%	0.001[c]
No	35	45%	26	87%	13	54%	
No, planning to test	24	69%	3	12%	5	38%	<0.001[d]
No, not planning to test	4	11%	17	65%	1	8%	
No, not sure if will test	7	20%	5	19%	7	54%	
I don't know	0	0%	0	0%	1	4%	
Have any of your other children had carrier testing?							
Yes	3	8%	1	6%	0	0%	
No	34	92%	15	88%	9	100%	
Other	0	0%	1	6%	0	0%	
Family Communication							
Have you told family members that they may be carriers?							
Yes	70	92%	9	32%	18	75%	<0.001[c]
No	6	8%	19	68%	6	25%	
No, we plan to tell the family	3	50%	1	5%	1	17%	
No, we do not plan to tell	0	0%	11	57%	2	33%	
No, I'm not sure whether we will tell family members	2	33%	4	21%	3	50	
No, other	1	17%	3	16%	0	0%	
Family Planning							
Will what you have learned influence family planning?							
No	28	38%	14	54%	9	38%	0.488[d]
Yes	26	35%	7	27%	6	25%	

	Carrier status of infant						
	Carrier (n=77)[a]		Non-Carrier (n=30)[a]		Other* (n=24)[a]		p-value
	n	%	n	%	n	%	
Yes, will have more children	18	69%	2	29%	2	33%	
Yes, won't have more children	5	19%	4	57%	2	33%	
Yes, other	3	12%	1	14%	2	33%	
Unsure	15	20%	4	15%	7	29%	
Other	5	7%	1	4%	2	8%	

[a]Missing data excluded;

[c]Chi-square *p*-value;

[d]Fisher's exact *p*-value

*other refers to mothers who responded 'don't know' or 'other' to the question regarding their infants' carrier status.