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SHORT COMMUNICATION

Interpretation and management of genetic test results by Canadian family physicians: a multiple choice survey of performance

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Abstract
Family physicians (FPs) will encounter genetic concerns within community practice. To determine how FPs compare to genetic counselors (GCs), a cross-sectional survey was distributed to Canadian FPs and GCs in 2019. The survey assessed risk analysis, counseling, and management of genetic information. FPs performed less well than GCs on each survey question and scenario ($p < 0.05$). Average overall survey scores for FPs were lower than GCs (62% vs. 93%, $p < 0.001$). Additional genetic training for FPs may help avoid potential harm.

Keywords Diagnostic tests · Evidence-based medicine · Family health · Genetics/genomics · Primary care · Survey research

Background

Rare single-gene disorders at birth have been estimated to affect approximately 1% of individuals worldwide (Blencowe et al. 2018). This is likely an underestimate of the true prevalence of genetic conditions, as it does not include adult-onset genetic conditions, common single-gene disorders (e.g., hemoglobin disorders, cystic fibrosis), or chromosome disorders. Furthermore, studies based on exome data have shown that each individual is a potential carrier of 11 recessive genetic disorders (Gambin et al. 2015). Given this, it should be expected that genetic concerns will be encountered in the practice of family medicine.

Canadian family medicine residents and practicing family physicians (FPs) recognize that they have knowledge gaps in the field of genetics, viewed in part to be due to the lack of relevant training in medical school and residency (Telner et al. 2008; Amara et al. 2018; Bonter et al. 2011). This translates to perceived difficulty managing genetic information (Amara et al. 2018; Bonter et al. 2011; Blaine et al. 2008; Carroll et al. 2016). Despite this, Canadian primary care providers endorse the importance of genetics within primary care and believe it will become more prevalent in the future (Telner et al. 2008; Carroll et al. 2016; Harding et al. 2019a). The strength of the patient-provider relationship in family medicine and knowledge of patients' medical and personal histories are important reasons for FP involvement in genetics (Carroll et al. 2016). Literature shows Canadian FPs support a role in providing genetic services, particularly with assessing family histories and making appropriate referrals (Telner et al. 2008; Carroll et al. 2016; Harding et al. 2019a). To date, the role the FPs will play in ordering genetic testing remains ill-defined. The majority of Canadian FPs report they are unable to interpret genetic test results; however, FPs are beginning to incorporate genetic testing into their practices (Bonter et al. 2011; Ronquillo et al. 2012; Christian S, Personal Communication. 2019).

Few studies have explored how FPs perform when assessing genetic information. Two American studies showed poor performance in assessing genetic risk (White et al. 2008; Bellcross et al. 2011). Canadian studies showed mixed performance (Carroll et al. 2011; Kadaoui et al. 2012; Carroll et al. 2009), with as high as 90% of survey respondents correctly assessing a hypothetical scenario on inherited risk (Carroll et al. 2009). Few studies to date have looked at how FPs perform when given actual genetic testing results. A small study explored management strategies of FPs for generally healthy patients with genomic results, but the physicians had previously received continuing medical education (CME)

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covering medical genetics (Vassy et al. 2018). The objective of our study was to compare the performance of Canadian FPs to Canadian GCs regarding interpretation and management of genetic testing results.

Methods

This study utilized an anonymous web-based cross-sectional survey.

Survey design

Demographic data, including years in practice, population of practice setting, province of practice, and self-rated knowledge of genetics, was collected on FPs. This was not collected on genetic counselors, as individual demographic factors would not be expected to impact their performance given their training in human genetics. Aside from the demographic questions, the survey was presented identically to FPs and GCs. It consisted of 4 scenarios—each composed of a written case history, genetic pedigree, and a genetic testing report from an accredited Canadian publicly run molecular diagnostics lab. Scenarios encompassed different genetic inheritance patterns and different genetic tests, including scenarios based around fragile X (X-linked inheritance), cystic fibrosis (autosomal recessive inheritance), hereditary breast and ovarian cancer (autosomal dominant inheritance), and undifferentiated developmental delay. Respondents answered a total of 10 questions (2–3 questions per scenario) regarding risk analysis, counseling, and management of the genetic information. The survey was reviewed and piloted on both FPs and GCs. The survey was reviewed by content experts for face validity and to ensure agreement on correct responses. The survey is available in the Supplementary information.

Study population and recruitment

FPs in Saskatchewan were recruited by email invitation via the Saskatchewan Medical Association in August 2019. Alberta FPs were recruited through an Alberta Medical Association email newsletter in early September 2019. Similarly, Canadian genetic counselors (GCs) were recruited via the Canadian Association of Genetic Counsellors listserv starting mid-August 2019. The survey closed to both groups after a 4–8-week period.

Data analysis

Data are expressed as means or counts (percentages). Responses were dichotomized (right or wrong) before analysis. Inter-group comparisons were performed using SPSS version 22 and Mann-Whitney U, Fisher's exact, or χ^2 tests, as appropriate. Differences were considered statistically significant at $p < 0.05$.

Results

A total of 172 respondents completed the survey. We do not have a defined number of FP recipients due to the newsletter recruitment method, so response rates among FPs could not be calculated. Of 124 FPs who started the survey, only 75 (60%) completed it. Eight completed surveys were excluded because the respondents were trainees, practiced outside of Saskatchewan or Alberta, or were not currently practicing. A total of 104 GCs started the survey and 97 (93%) completed it. Of the GCs, 92% currently work in a GC role. The remaining 8% of GC respondents included 1 retired GC, and 4 respondents who were working in industry and academic roles. There were 330 registered GCs in Canada at the time of survey distribution for a response rate among GCs of 29%. Demographic information of FP respondents is presented in Table 1.

GCs were significantly more likely to correctly answer each of the individual questions than FPs (Table 2). The overall average survey score for GCs was 93%, whereas FPs averaged 62%. Similarly, GCs outperformed FPs on each of the scenarios (Table 3). The performance on individual questions, each scenario, and overall survey score for GCs was significantly higher than that of FPs. None of the FPs responded correctly to all questions. FPs with ≤10 year of experience

Table 1 Demographic characteristics of the 67 Canadian family physicians who provided responses to the genetics knowledge questionnaire in 2019

	n (%)
Years in practice	
<5	19 (28.4%)
5–10	11 (16.4%)
11–19	21 (31.3%)
20+	16 (23.9%)
Practice setting	
Urban	41 (61.2%)
Regional	9 (13.4%)
Rural/remote	17 (25.4%)
Province of practice	
Alberta	28 (41.8%)
Saskatchewan	39 (58.2%)
Self-perceived genetic knowledge	
Very poor	2 (3.0%)
Poor	19 (28.4%)
Average	44 (65.7%)
Above average	2 (3.0%)
Excellent	0 (0.0%)

Table 2 Comparison of question-by-question performance of the 67 Canadian family physicians and 97 Canadian genetic counselors within each of the genetic testing scenario assessed in 2019. Values represent frequency and percentage of family physicians and genetic counselors who correctly answering each of the questions

Scenario description	Question theme	Correct responses by group (*n*/%)		*p* value
		Family physicians (*n* = 67)	Genetic counselor (*n* = 97)	
X-linked inheritance and fragile X testing	Likelihood of inheritance in males	49 (73%)	90 (93%)	0.001
	Likelihood of inheritance in females	47 (70%)	97 (100%)	<0.001
	Appropriate follow-up	59 (88%)	95 (98%)	0.009
Autosomal recessive inheritance and cystic fibrosis testing	Assess carrier risk	35 (53%)	89 (92%)	<0.001
	Likelihood of affected offspring	50 (75%)	91 (94%)	0.001
	Appropriate follow-up testing	14 (21%)	68 (70%)	<0.001
Autosomal dominant inheritance and hereditary breast and ovarian cancer (BRCA 1/2)	Appropriate testing and risk stratification	37 (55%)	94 (97%)	<0.001
	Results interpretation and management	49 (73%)	89 (92%)	0.001
Undifferentiated developmental delay and microarray testing	Appropriateness of testing in relatives	56 (84%)	97 (100%)	<0.001
	Risk assessment and follow-up plans	22 (33%)	95 (98%)	<0.001

did not show any statistically significant differences when compared to physicians with >10 years of experience (Supplementary information, Tables S1 and S2)

Discussion

This is one of the first studies assessing how FPs perform when presented with familial genetic information and actual genetic test results. As such, no validated instruments exist that could be used for this study. However, GCs would be expected to perform very well on a survey with clear and well-written case histories and multiple-choice responses. The data suggest that that the survey was well-written as ≥ 92% of GCs selected the correct response for 9 out of the 10 survey questions.

FPs performed worse than GCs on overall survey performance, scenario-by-scenario performance, and question-by-question performance. Based on the discrepancy in genetic training between the groups, this is not surprising. However, these scenarios reflect true-to-life scenarios that could present to the FPs office. The data implies that FPs are more likely to misinterpret or mismanage basic genetic information. For example, 49% of FPs were unable to correctly estimate carrier status for an autosomal recessive condition, although they tended to err on the side of overestimating risk in this scenario. The poorest performance of FPs was on the scenario regarding microarray testing in a patient with undifferentiated developmental delay. Microarray testing has replaced karyotype as a first-line genetic test and is becoming available for FPs to order in some settings. In this scenario, 69% of the responses to the scenario were inappropriate, highlighting an important need for additional training on this technology.

Table 3 Comparison of performance of Canadian 67 family physicians and 97 genetic counselors. FPs overall and for each of the genetic testing scenario

Scenario	Scenario description	Mean % correct		Absolute % difference	*p* value
		FPs	GCs		
1	X-linked inheritance, fragile X testing	77	95	18	<0.001*
2	Autosomal recessive inheritance, cystic fibrosis testing	49	85	36	<0.001*
3	Autosomal dominant inheritance, hereditary breast and ovarian cancer (BRCA 1/2) testing	64	95	31	<0.001*
4	Undifferentiated developmental delay, microarray testing	58	99	42	<0.001*
	Overall	62	93	31	<0.001*

*Indicates statistical significance at $p < 0.05$ using Mann-Whitney *U* analysis

Mismanagement of genetic information can result in harm to patients or their families. The goal of this survey was to specifically assess genetic risk assessment and management by FPs, and therefore did not give the option of referral to Medical Genetics. In practice, FPs uncomfortable with interpreting or managing genetic information would likely refer; however, FPs have also indicated that they need additional education to refer appropriately (Harding et al. 2019a, b). Inappropriate referrals are associated with poor patient outcomes and increased cost (Greenwood-Lee et al. 2018).

Our study had several limitations. Due to the recruitment methods, we do not have an accurate estimate of the response rate. Thus, the survey may be prone to non-response bias. However, given similarities in accreditation standards in medical training, physicians are likely more similar regarding their knowledge and behavior than the general population. Therefore, physician surveys may be less prone to non-response bias than surveys of other groups (Kellerman and Herold 2001). If our study suffers from non-response bias, it is likely that the FP group was biased in a positive direction. Almost 40% of FPs who started the survey did not complete it, possibly because the scenarios were felt to be too complex for their level of genetic training. For example, one respondent commented "I did not answer any of your questions as I do not know the answers. Not sure why you asked us to do this complex questionnaire other than to insult us?" True performance of FPs as a group, could therefore be worse than indicated by the current study. Additional limitations of this study include the lack of a validated instrument, voluntary participation, and limited geographic scope.

A number of groups have suggested or studied core genetic competencies for non-geneticist physicians, including the American Association of Family Physicians (2019) and others (National Coalition for Health Professional Education in Genetics 2007; Tognetto et al. 2019). The scenarios presented in this study evaluate many of the core competencies suggested by these various groups, including understanding inheritance patterns, interpretation of multigenerational histories, and understanding indications for genetic testing. Modes of inheritance were explicitly stated in the scenarios to minimize the need for condition-specific knowledge.

Not only does this study assess FP performance with core genetic concepts, but it may also aid in determining FP readiness to take on a greater role in offering genetic testing within the era of genomics. A number of Canadian provinces have reported wait times to access genetic services of >2 years, in some cases resulting in under-referral of patients who would be eligible for genetic testing (Alberta Health Services 2020; Cancer Care Ontario 2018). Given this, it is possible that in the coming years, there may be a shift that leaves FPs increasingly responsible for more "routine" genetic testing, leaving more complex cases to genetic specialists. This study suggests that as a group, FPs are not well-equipped to take on this task at the primary care level with the current standards of genetic training in medical school and residency.

Education may enhance genetics knowledge in primary care (Telner et al. 2017). There has been an increasing number of publications in recent years exploring genomics training for non-genetic health professionals (Talwar et al. 2017). Most schools teach medical genetics during the first 2 years of undergraduate medicine (Plunkett-Rondeau et al. 2015). Rapid changes in genomics renders much of what trainees learn out of date by the time they enter practice (Korf et al. 2014). Similar to Carroll et al. (2019), we did not see differences in the knowledge of physicians with regards to year in practice. Thus, education targeting learners late in training and CME is needed. Our results indicate that patterns of inheritance, risk assessment, and understanding the benefits, risks, and limitations of genetic testing need to be included in curriculums.

Education alone is not effective in optimizing clinical care by health professionals (Clyman et al. 2007). Other strategies and research beyond education are required to improve FPs' ability to interpret and manage genetic testing. Collaboration between laboratories, GCs, medical geneticists, and FPs in the co-development of guidelines and clinical tools might facilitate the effective use of genetic testing by primary care providers.

Conclusions

Misinterpretation or mismanagement of genetic information can have a marked clinical impact on patients, their children, or their families. Given the disparity in genetic training, it is not surprising that GCs performed significantly better than FPs on all 4 scenarios presented in this survey and on both individual question and overall test performance. Given the strain on genetic services, it is quite possible that FPs may become increasingly responsible for assessing, interpreting, and managing genetic information in the years to come. With current levels of training, FPs do not appear to be adequately prepared to fill this role. Additional training at the residency and CME levels may help to fill this knowledge gap in the genomic era.

Supplementary Information The online version contains supplementary material available at https://doi.org/10.1007/s12687-021-00511-w.

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Author contribution Michelle McCarron, Stephanie Skinner, and Sarah Liskowich contributed to the study conception and design. Data analysis was performed by Michelle McCarron and Adam Clay. The first draft of the manuscript was written by Stephanie Skinner, and all authors commented on previous versions of the manuscript. All authors read and approved the final manuscript.

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Data availability Due to the nature of this research, participants of this study did not agree for their data to be shared publicly, so supporting data is not available.

Code availability Not applicable

Declarations

Ethics approval Ethics approval was obtained from the Research Ethics Board of the former Regina Qu'Appelle Health Authority (REB-19-25).

Consent to participate Informed consent was obtained from all individual participants included in the study.

Conflict of interest The authors declare no competing interests.

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