

# **Genetics in Medicine**

## **Polygenic Risk Scores and Risk-Stratified Breast Cancer Screening: Familiarity and Perspectives of Healthcare Professionals**

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# **Polygenic Risk Scores and Risk-Stratified Breast Cancer Screening: Familiarity and Perspectives of Healthcare Professionals**

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**Abstract:**

**Purpose:** Healthcare professionals are expected to take on an active role in the implementation of risk-based cancer prevention strategies. This study aims to explore healthcare professionals’ i) self-reported familiarity with the concept of polygenic risk scores (PRS), ii) perceived level of knowledge regarding risk-stratified breast cancer (BC) screening, and iii) preferences for continuing professional development (CPD).

**Methods:** A cross-sectional survey using a bilingual – English/French – online questionnaire disseminated by healthcare professional associations across Canada between November 2020 and May 2021.

**Results:** A total of 593 professionals completed more than two items and 453 responded to all questions. A total of 432 (94%) participants were female, 103 (22%) were physicians and 323 (70%) were nurses. Participants reported to be unfamiliar (20%), very unfamiliar (32%) with, or did not know (41%) the concept of PRS. The majority of participants reported not having enough knowledge about risk-stratified BC screening (61%) and that they would require more training (77%). Online courses and webinar conferences were the preferred CPD modalities.

**Conclusion:** The study indicates that healthcare professionals are currently not familiar with the concept of PRS or a risk-stratified approach to BC screening. Online information and training seem to be an essential knowledge transfer modality.

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## 1. Introduction

Breast cancer (BC) remains the most common cancer diagnosed among women worldwide.<sup>1</sup> There is compelling evidence suggesting that early detection of BC significantly reduces mortality from the disease<sup>2</sup>, but this comes with risks of false positive screening results, overdiagnosis, and psychological impacts. Emerging evidence suggests that a risk-stratified approach to BC screening can improve its benefit-risk ratio by targeting those women most likely to benefit from it, potentially leading to reduced BC-specific mortality as well as allowing for more efficient allocation of health care resources.<sup>4</sup> This stratification approach, currently under investigation in Canada<sup>5</sup>, the United-States (US)<sup>6</sup> and in Europe<sup>7</sup>, encompasses three steps: first, collection of women’s personal and genetic information; second, calculation of their risk of developing BC within a given time horizon using a risk prediction model; third, disclosure of the risk level and the possible screening and risk reduction actions to participants. Several BC risk prediction models are now incorporating a polygenic risk score (PRS).<sup>8</sup> The PRS – derived from genome-wide associations studies (GWAS) – is a score that combines the effects of several common genetic variants with small individual effect sizes, but when combined are strongly associated with the risk of developing the medical condition.<sup>9</sup>

The integration of risk-stratified BC screening into health systems will require healthcare

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professionals (HCPs) to demonstrate new competencies in terms of knowledge, skills and attitudes. For example, primary care professionals and those from medical specialties other than genetics could be expected to explain both the harms and benefits of risk-stratified BC screening, interpret and communicate to patients their risk level obtained through a risk prediction model and advise them on screening and preventive strategies.<sup>10,11</sup> Some of the information to be exchanged through this process is complex. An example is the explanation of the calculation of the PRS which requires a good level of familiarity, and ideally knowledge, for its responsible integration to clinical practice .

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However, little is known regarding HCPs' familiarity with the concept of PRS and their perspectives regarding risk-stratified BC screening. Two smaller-size studies surveyed HCPs' familiarity and use of PRS<sup>13,14</sup>, but over 84% of the participants were genetic counselors. While genetic counselors are an important professional group to consider for the dissemination and implementation of risk-stratified screening approach, other HCPs, such as those involved in primary care, also need to be considered. Also, investigation efforts to collect Canadian HCPs' perspectives regarding risk-stratified BC screening have so far been mainly conducted through qualitative methodologies.<sup>15,16</sup> A quantitative survey on a

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384 larger sample of HCPs from different medical specialties is thus needed in order to appraise the level of  
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585 training required to support optimal implementation in the health care system.  
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886 Canada has a universal healthcare system that emphasizes public administration, comprehensiveness,  
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1087 universality, portability and accessibility (Canada Health Act). Each of its jurisdictions (i.e. 10 provinces  
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1288 and 3 territories) determine what medical acts are covered within their healthcare plan . With the  
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1590 include offering regular BC screening mammograms<sup>18</sup>. Some provinces, such as Ontario and British  
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1791 Columbia, also have High Risk programs offering genetics counseling, testing and/or enhanced screening  
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1992 strategies to women at increased risk . In all instances, HCPs in the primary care settings (nurses  
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2193 practitioners and family physicians alike) are advised to routinely address BC screening practices with  
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2394 their patients .  
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2695 This study aims to explore HCPs' i) self-reported familiarity with concept of PRS, ii) perceived level  
2796 of knowledge regarding risk-stratified BC screening, and iii) preferences for continuing professional  
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2997 development (CPD). Evidence generated by this study will provide crucial information about current  
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3198 professionals' appraisal of their knowledge. This will support the design of CPD aiming to develop  
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3399 competency in supporting patients in understanding their BC risk level, making informed decisions  
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3500 related to screening and preventive interventions, and potentially avoiding unnecessary adverse  
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3701 psychosocial impacts.  
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## 4103 2. Materials and Methods 39

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4304 This study is part of PERSPECTIVE I&I (Personalized risk assessment for prevention and early detection  
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4505 of breast cancer: Integration and Implementation), a major Canadian initiative assessing the feasibility  
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and acceptability of implementing a risk-stratified BC screening approach.<sup>5</sup>

## **2.1. Study Design and Participants**

A cross-sectional study was conducted using an anonymous self-administrated online questionnaire targeting all HCPs interested in providing their opinions, attitudes and expectations regarding risk-stratified BC screening. While there were no inclusion/exclusion criteria for participants, our promotion and diffusion strategy targeted physicians and nurses from all medical specialties. The study invitation with the link to the questionnaire was disseminated between November 2020 and May 2021 through

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several professional associations and healthcare institutions’ newsletters and communication platforms across Canada (see Supplementary Material S1) as well as through PERSPECTIVE I&I co-investigators’ networks. The first page of our questionnaire provided elements of context about the study and informed participants that consent was implied by the voluntary completion of the questionnaire. The CHU de Québec-Université Laval’s Institutional Review Board approved this study (registration number: F9-55772).

## 2.2. Questionnaire Development

The questionnaire was developed in French and English by a multidisciplinary team of clinicians, epidemiologists, and social scientists after reviewing the relevant literature.<sup>15,16,22-24</sup> The questionnaire had a total of 17 questions, with data from 10 analysed to achieve our three objectives (see Supplementary Material S2). After a short preamble explaining the risk-based BC screening approach, questions covered familiarity with the concept of PRS (1 question), opinions regarding their level of knowledge, the status of their training and the future professional curriculum on risk assessment, including genetic factors (1 question comprising 5 statements), preferences for continuing professional education (3 questions), and sociodemographic and professional status (5 questions). The French and English questionnaires were pilot-tested within the network of physicians collaborating on the study and comments were addressed by the research team. The REDCap platform was used for the questionnaire web-based interface.<sup>25</sup>

## 2.3. Statistical Analyses

The five-point Likert scale of the question assessing participants’ level of familiarity with the concept of PRS was categorized on three levels: “Very familiar and familiar”, “Very unfamiliar and unfamiliar” and “Don’t know this concept”. HCPs were categorized as “Physician”, “Nurse” or “Other”. Medical specialties were categorized as “Family medicine/Primary care”, “Oncology” and “Other”. The number of years of experience was categorized as follow: less than 5 years, between 5 and 14 years, between 15 and 25 years, and more than 25 years. The region of practice was categorized as “Province of Québec”, “Province of Ontario”, “Other Canadian provinces and territories”.

Descriptive statistics were used to summarize participant responses. Chi-square tests were used to explore whether participants’ level of familiarity with the concept of PRS differed according to sociodemographic and professional status variables. Dummy variables were created for missing responses. Analyses using listwise deletion of missing variables were also conducted as a sensitivity



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analysis<sup>26</sup>. All tests were two-sided with a 0.05 level of significance. All statistical analyses were performed using SAS software, Version 9.4 (Copyright © 2016 by SAS Institute Inc., Cary, NC, USA).

### 3. Results:

A total of 593 opened the survey link and completed more than two questions. A total of 453 participants responded to all questions. Overall, 432 (93.5%) participants were female, 103 (22.3%) were physicians, and 323 (69.7%) were nurses (i.e. nurses and nurse practitioners) (Table 1). The distribution by speciality was as follows: family medicine/primary care (36.1%), oncology (12.8%), and other (51.1%). Other medical specialties included: internal medicine, surgery, emergency, palliative care, public health medicine, radiology, and obstetrics – gynecology. The province of Québec was the most frequent region of practice for participants (82.9%), followed by Ontario (10.1%), and other Canadian provinces and territories (7.0%). Participants’ most frequent practice settings included academic hospital (28.9%), community hospital (21.3%), community health centre (17%), and family health team, group or network (16.3%). Finally, more than 89% of participants agreed or strongly agreed that breast cancer screening is an effective method for early detection of breast cancer.

**Table 1. Participants’ characteristics (N=593)**

The vast majority of participants reported to be unfamiliar (19.9%), very unfamiliar (31.9%) with or did not know (40.5%) the concept of PRS (Figure 1). Exploratory univariate analyses revealed that the profession, medical specialty, and region of practice were associated with a different report of familiarity with the concept of PRS with doctors being more familiar with the concept compared to other professions, oncologists reporting more familiarity than other medical specialties and people from the province of Québec reporting less familiarity with the concept of PRS. Gender, number of years of practice, and practice setting were not associated with familiarity with the concept of PRS (Figure 2 and Supplementary Material S3). Similar pattern of associations were observed when missing data were excluded.

**Figure 1. Participants’ level of familiarity with the concept of polygenic risk score (PRS) (N=593)**

**Figure 2. Association between familiarity with the concept of PRS and sociodemographic variables (N = 593)**

When asked about their opinion regarding their level of knowledge towards risk-stratified BC screening and the ideal future professional curriculum on risk assessment, including genetic factors, the

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vast majority of participants answered that: i) they do not have enough knowledge (60.5%), ii) they would require more training (76.9%), and iii) that the ideal medical and nursing curriculum should include more on this topic (70.3% and 71.3%, respectively) (Figure 3). However, only 45.9% answered that they would have time to educate themselves on risk-stratified BC screening

**Figure 3. Participants’ perspective regarding their education and continuous professional development (CPD) (N = 593)**

Figure 4 presents the preferred CPD resources, dissemination modalities and topics to include in educational resources. Higher participants’ preference was observed for online training specific to risk-stratified BC screening (26%) with topics addressing the basics of risk-stratified BC screening (16%) and its interpretation (15%). Participants were less interested by general information on genetics and the ethical, legal, and social challenges of risk-stratified BC screening.

**Figure 4. Participants’ preferred resources and CPD modalities in general (a), to learn more about risk-based breast cancer (BC) screening (b) and topics to be included in resource material (c) (participants invited to check all that apply)**

**4. Discussion**

This study provides important information on familiarity with the concept of PRS, perceived level of knowledge regarding risk-stratified BC screening, and preferences for CPD of HCPs not trained in genetics. Overall, HCPs reported low level of familiarity with the concept of PRS and limited knowledge regarding risk-stratified BC screening. The vast majority acknowledged their needs for CPD on these topics and would favor resources delivered online.

To our knowledge, only two smaller-size studies (i.e. sample sizes of 105<sup>14</sup> and 120<sup>13</sup>) have reported HCPs’ level of familiarity with the concept of PRS. However, both studies were focused primarily on genetic counselors. Thus, the results of these studies are not comparable with ours since our study population was composed mainly of professionals not trained in genetics. Our study complements the evidence generated by these previous studies by providing the perspectives of a diverse group of HCPs and by highlighting the fact that, unlike genetic counselors, professionals not trained in genetics currently report a low level of familiarity with concept of PRS. Having basic knowledge regarding the calculation and implications of a PRS is important for several HCPs, including front line professionals such as nurses and primary care physicians. Indeed, if calculations of PRS are implemented in clinical practice, they would need to answer questions related to PRS results and support their patients in their

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203 decision process regarding appropriate screening recommendations and preventative options. HCPs  
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1574 require more training (76.9%), and iii) that the ideal medical and nursing curriculum should include more  
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1775 on this topic (70.3% and 71.3%, respectively) (Figure 3). However, only 45.9% answered that they would  
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should also be knowledgeable of the potential limitations of PRS and be able to convey a balanced message to their patients.<sup>27</sup> Finally, according to different possible implementation scenarios, front line HCPs may have an important role in identifying and referring individuals for whom a risk assessment that included a PRS calculation is most indicated.<sup>15,28</sup>

The observation that the vast majority of our participants stated that they do not have enough knowledge about risk-stratified BC screening and would require more training is in line with several studies reporting that HCPs feel unprepared and lack the appropriate knowledge to competently integrate emerging genomic information into their practice.<sup>14-16,29-32</sup> Scientific literature about the concept of PRS and risk-stratified BC screening has been published since at least 2015.<sup>33,34</sup> Thus, reported low level of familiarity and knowledge of HCPs about these two aspects suggests that active dissemination strategies are required.

To ensure a successful integration of the PRS and risk-stratified screening approaches, a comprehensive portfolio of CPD activities – adapted to the different professional groups and medical specialties – is necessary. Academic institutions will probably need to adapt their curriculum to address these knowledge gaps, and authoritative associations should be called upon to provide point-of-care resources, clinical guidelines, and implementation protocols for the responsible use of PRS information and sound implementation of risk-stratified BC screening.<sup>12</sup>

Although these analyses were exploratory, it is interesting to note significant differences observed on the level of familiarity with the concept of PRS according to participants’ profession, medical specialty, and region of practice. Previous studies assessing level of knowledge with genetics and/or genomics have reported similar differences across professions<sup>30,35,36</sup>, medical specialties<sup>29,37,38</sup> and geographical locations. These differences on the level of familiarity with the concept of PRS are

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ervices professionals are offering as well as their exposure to genetic services within their health care  
institutions.<sup>36</sup> Knowledgeable of the potential limitations of PRS and has implemented a High Risk  
Message Board for patients.<sup>27</sup> Finally, a thorough review of possible implementation issues for HCPs, front lines  
related to genetics and genomics risk of BC. Uncovering professional group differences on the familiarity  
and knowledge about the concept of PRS and risk- stratified BC screening should be explored further  
through a more comprehensive assessment among different HCPs. This could serve as a parameter of  
guidance for the development of tailored CPD activities and resources.

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Our participants’ preference for online CPD resources is in line with other studies<sup>14,29,41</sup>, although the in-person CPD modality was preferred in one.<sup>42</sup> We may hypothesize that HCPs’ preference for the online modality has probably increased since the occurrence of the Covid-19 pandemic. Online resources have the advantage of being available “just in time”, exactly when HCPs need it and are ready to integrate this in their practice and skill set. This immediacy feature is coherent with an important adult learning theory principle which suggests that learners are interested in acquiring knowledge that ~~have~~has immediate relevance and impact.<sup>43</sup>

#### 4.1. Strengths and Limitations

To our knowledge, the sample size of this study is the largest and includes the most diversified population of HCPs to date to characterize their level of familiarity and perspectives on the concept of PRS and the risk-stratified BC screening approach. The recruitment method was multifaceted and primarily through professional associations and healthcare institutions. Such a recruitment scale is particularly impressive in the context of the Covid-19 pandemic.

Nevertheless, it is important to recognize the limitations of our study. While our questionnaire was designed to be of 15-minute duration in order to accommodate a target population with many competing priorities, a more detailed knowledge status and learning needs collection tool would be required as this is a crucial step in establishing sound CPD curriculum.<sup>44</sup> It is important to recognize that an online recruitment and data collection strategy may result in a greater proportion of participants preferring electronic CPD. Our sample distribution is not representative of the Canadian healthcare professional population. Supplementary material S4 contrasts the demographics of participants with that of the Canadian physicians and nurses workforce. Our sample has a higher proportion of female and of

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professionals from the province of Québec compared to the National statistics . However, the proportion

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2 ed family medicine as their medical specialty is comparable to the national statistics.

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3 Our participants' preference for online CPD resources is in line with other studies<sup>14,29,41</sup>, although  
4 We believe though that our results offer an indication that professionals not trained in genetics  
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real level of familiarity with the concept of PRS and knowledge of risk-stratified BC screening of HCPs is even lower than what we observed.

### 5. Conclusion

Current use of PRS testing is at an early stage of integration.<sup>12</sup> While the risk-stratified BC screening based on information such as the PRS is not yet part of any Canadian provinces or territories' public health measures, it is currently undergoing effectiveness-implementation studies in Canada<sup>5</sup>, as well as in the U.S.<sup>6</sup> and in Europe<sup>7</sup>. There is therefore a window of opportunity for professional associations, healthcare institutions and public health or government agencies overseeing screening programs to proactively plan for knowledge dissemination strategies that will effectively support HCPs involved at different contact points in the integration of this emerging genomics strategy. The fact that a lack of knowledge has been identified as the most frequent barrier to the implementation of genetics and genomics in practices justifies the relevance of investing in workforce preparation and CPD activities tailored to healthcare professionals existing knowledge and clinical practice needs.

### Data Availability

Additional data are available from the corresponding author upon request.

### Acknowledgements

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### Author Contributions

Supervision: HN; Writing-original draft: JL, ACB, CMB, HN; Writing-review & editing: All authors; Conceptualization: All authors; Data curation: JL, ACB, CMB; Formal analysis: JL, ACB, CMB, HN;

### Ethics Declaration

Project administration: JL, HN; Resources: The study was approved by the Institutional Review Board of: CHU de Québec–Université Laval (2021-5136). Informed consent was obtained from all participants when they opened the survey link.



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Data were collected in a completely anonymous fashion - the information never had identifiers associated with it .

**Conflict of Interest**

The authors declare no conflict of interest. The funders had no role in the design of the study; in the collection, analyses, or interpretation of data; in the writing of the manuscript, or in the decision to publish the results.

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# **Polygenic Risk Scores and Risk-Stratified Breast Cancer Screening: Familiarity and Perspectives of Healthcare Professionals**

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**Abstract:**

**Purpose:** Healthcare professionals are expected to take on an active role in the implementation of risk-based cancer prevention strategies. This study aims to explore healthcare professionals’ i) self-reported familiarity with the concept of polygenic risk scores (PRS), ii) perceived level of knowledge regarding risk-stratified breast cancer (BC) screening, and iii) preferences for continuing professional development (CPD).

**Methods:** A cross-sectional survey using a bilingual – English/French – online questionnaire disseminated by healthcare professional associations across Canada between November 2020 and May 2021.

**Results:** A total of 593 professionals completed more than two items and 453 responded to all questions. A total of 432 (94%) participants were female, 103 (22%) were physicians and 323 (70%) were nurses. Participants reported to be unfamiliar (20%), very unfamiliar (32%) with, or did not know (41%) the concept of PRS. The majority of participants reported not having enough knowledge about risk-stratified BC screening (61%) and that they would require more training (77%). Online courses and webinar conferences were the preferred CPD modalities.

**Conclusion:** The study indicates that healthcare professionals are currently not familiar with the concept of PRS or a risk-stratified approach to BC screening. Online information and training seem to be an essential knowledge transfer modality.

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## 1. Introduction

Breast cancer (BC) remains the most common cancer diagnosed among women worldwide.<sup>1</sup> There is compelling evidence suggesting that early detection of BC significantly reduces mortality from the disease<sup>2</sup>, but this comes with risks of false positive screening results, overdiagnosis, and psychological impacts. Emerging evidence suggests that a risk-stratified approach to BC screening can improve its benefit-risk ratio by targeting those women most likely to benefit from it, potentially leading to reduced BC-specific mortality as well as allowing for more efficient allocation of health care resources.<sup>4</sup> This stratification approach, currently under investigation in Canada<sup>5</sup>, the United-States (US)<sup>6</sup> and in Europe<sup>7</sup>, encompasses three steps: first, collection of women’s personal and genetic information; second, calculation of their risk of developing BC within a given time horizon using a risk prediction model; third, disclosure of the risk level and the possible screening and risk reduction actions to participants. Several BC risk prediction models are now incorporating a polygenic risk score (PRS).<sup>8</sup> The PRS – derived from genome-wide associations studies (GWAS) – is a score that combines the effects of several common genetic variants with small individual effect sizes, but when combined are strongly associated with the risk of developing the medical condition.<sup>9</sup>

The integration of risk-stratified BC screening into health systems will require healthcare

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professionals (HCPs) to demonstrate new competencies in terms of knowledge, skills and attitudes. For example, primary care professionals and those from medical specialties other than genetics could be expected to explain both the harms and benefits of risk-stratified BC screening, interpret and communicate to patients their risk level obtained through a risk prediction model and advise them on screening and preventive strategies.<sup>10,11</sup> Some of the information to be exchanged through this process is complex. An example is the explanation of the calculation of the PRS which requires a good level of familiarity, and ideally knowledge, for its responsible integration to clinical practice .

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However, little is known regarding HCPs' familiarity with the concept of PRS and their perspectives regarding risk-stratified BC screening. Two smaller-size studies surveyed HCPs' familiarity and use of PRS<sup>13,14</sup>, but over 84% of the participants were genetic counselors. While genetic counselors are an important professional group to consider for the dissemination and implementation of risk-stratified screening approach, other HCPs, such as those involved in primary care, also need to be considered. Also, investigation efforts to collect Canadian HCPs' perspectives regarding risk-stratified BC screening have so far been mainly conducted through qualitative methodologies.<sup>15,16</sup> A quantitative survey on a

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384 larger sample of HCPs from different medical specialties is thus needed in order to appraise the level of  
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585 training required to support optimal implementation in the health care system.  
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886 Canada has a universal healthcare system that emphasizes public administration, comprehensiveness,  
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1087 universality, portability and accessibility (Canada Health Act). Each of its jurisdictions (i.e. 10 provinces  
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1288 and 3 territories) determine what medical acts are covered within their healthcare plan . With the  
1389 exception of one territory, all jurisdictions also implement an organized public health program that  
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1590 include offering regular BC screening mammograms<sup>18</sup>. Some provinces, such as Ontario and British  
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1791 Columbia, also have High Risk programs offering genetics counseling, testing and/or enhanced screening  
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1992 strategies to women at increased risk . In all instances, HCPs in the primary care settings (nurses  
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2193 practitioners and family physicians alike) are advised to routinely address BC screening practices with  
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2695 This study aims to explore HCPs' i) self-reported familiarity with concept of PRS, ii) perceived level  
2796 of knowledge regarding risk-stratified BC screening, and iii) preferences for continuing professional  
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2997 development (CPD). Evidence generated by this study will provide crucial information about current  
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3198 professionals' appraisal of their knowledge. This will support the design of CPD aiming to develop  
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3399 competency in supporting patients in understanding their BC risk level, making informed decisions  
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3500 related to screening and preventive interventions, and potentially avoiding unnecessary adverse  
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## 4103 2. Materials and Methods 39

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4304 This study is part of PERSPECTIVE I&I (Personalized risk assessment for prevention and early detection  
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4505 of breast cancer: Integration and Implementation), a major Canadian initiative assessing the feasibility  
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and acceptability of implementing a risk-stratified BC screening approach.<sup>5</sup>

## **2.1. Study Design and Participants**

A cross-sectional study was conducted using an anonymous self-administrated online questionnaire targeting all HCPs interested in providing their opinions, attitudes and expectations regarding risk-stratified BC screening. While there were no inclusion/exclusion criteria for participants, our promotion and diffusion strategy targeted physicians and nurses from all medical specialties. The study invitation with the link to the questionnaire was disseminated between November 2020 and May 2021 through

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several professional associations and healthcare institutions’ newsletters and communication platforms across Canada (see Supplementary Material S1) as well as through PERSPECTIVE I&I co-investigators’ networks. The first page of our questionnaire provided elements of context about the study and informed participants that consent was implied by the voluntary completion of the questionnaire. The CHU de Québec-Université Laval’s Institutional Review Board approved this study (registration number: F9-55772).

## 2.2. Questionnaire Development

The questionnaire was developed in French and English by a multidisciplinary team of clinicians, epidemiologists, and social scientists after reviewing the relevant literature.<sup>15,16,22-24</sup> The questionnaire had a total of 17 questions, with data from 10 analysed to achieve our three objectives (see Supplementary Material S2). After a short preamble explaining the risk-based BC screening approach, questions covered familiarity with the concept of PRS (1 question), opinions regarding their level of knowledge, the status of their training and the future professional curriculum on risk assessment, including genetic factors (1 question comprising 5 statements), preferences for continuing professional education (3 questions), and sociodemographic and professional status (5 questions). The French and English questionnaires were pilot-tested within the network of physicians collaborating on the study and comments were addressed by the research team. The REDCap platform was used for the questionnaire web-based interface.<sup>25</sup>

## 2.3. Statistical Analyses

The five-point Likert scale of the question assessing participants’ level of familiarity with the concept of PRS was categorized on three levels: “Very familiar and familiar”, “Very unfamiliar and unfamiliar” and “Don’t know this concept”. HCPs were categorized as “Physician”, “Nurse” or “Other”. Medical specialties were categorized as “Family medicine/Primary care”, “Oncology” and “Other”. The number of years of experience was categorized as follow: less than 5 years, between 5 and 14 years, between 15 and 25 years, and more than 25 years. The region of practice was categorized as “Province of Québec”, “Province of Ontario”, “Other Canadian provinces and territories”.

Descriptive statistics were used to summarize participant responses. Chi-square tests were used to explore whether participants’ level of familiarity with the concept of PRS differed according to sociodemographic and professional status variables. Dummy variables were created for missing responses. Analyses using listwise deletion of missing variables were also conducted as a sensitivity

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analysis<sup>26</sup>. All tests were two-sided with a 0.05 level of significance. All statistical analyses were performed using SAS software, Version 9.4 (Copyright © 2016 by SAS Institute Inc., Cary, NC, USA).

**3. Results:**

A total of 593 opened the survey link and completed more than two questions. A total of 453 participants responded to all questions. Overall, 432 (93.5%) participants were female, 103 (22.3%) were physicians, and 323 (69.7%) were nurses (i.e. nurses and nurse practitioners) (Table 1). The distribution by speciality was as follows: family medicine/primary care (36.1%), oncology (12.8%), and other (51.1%). Other medical specialties included: internal medicine, surgery, emergency, palliative care, public health medicine, radiology, and obstetrics – gynecology. The province of Québec was the most frequent region of practice for participants (82.9%), followed by Ontario (10.1%), and other Canadian provinces and territories (7.0%). Participants’ most frequent practice settings included academic hospital (28.9%), community hospital (21.3%), community health centre (17%), and family health team, group or network (16.3%). Finally, more than 89% of participants agreed or strongly agreed that breast cancer screening is an effective method for early detection of breast cancer.

**Table 1. Participants’ characteristics (N=593)**

The vast majority of participants reported to be unfamiliar (19.9%), very unfamiliar (31.9%) with or did not know (40.5%) the concept of PRS (Figure 1). Exploratory univariate analyses revealed that the profession, medical specialty, and region of practice were associated with a different report of familiarity with the concept of PRS with doctors being more familiar with the concept compared to other professions, oncologists reporting more familiarity than other medical specialties and people from the province of Québec reporting less familiarity with the concept of PRS. Gender, number of years of practice, and practice setting were not associated with familiarity with the concept of PRS (Figure 2 and Supplementary Material S3). Similar pattern of associations were observed when missing data were excluded.

**Figure 1. Participants’ level of familiarity with the concept of polygenic risk score (PRS) (N=593)**

**Figure 2. Association between familiarity with the concept of PRS and sociodemographic variables (N = 593)**

When asked about their opinion regarding their level of knowledge towards risk-stratified BC screening and the ideal future professional curriculum on risk assessment, including genetic factors, the

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vast majority of participants answered that: i) they do not have enough knowledge (60.5%), ii) they would require more training (76.9%), and iii) that the ideal medical and nursing curriculum should include more on this topic (70.3% and 71.3%, respectively) (Figure 3). However, only 45.9% answered that they would have time to educate themselves on risk-stratified BC screening

**Figure 3. Participants’ perspective regarding their education and continuous professional development (CPD) (N = 593)**

Figure 4 presents the preferred CPD resources, dissemination modalities and topics to include in educational resources. Higher participants’ preference was observed for online training specific to risk-stratified BC screening (26%) with topics addressing the basics of risk-stratified BC screening (16%) and its interpretation (15%). Participants were less interested by general information on genetics and the ethical, legal, and social challenges of risk-stratified BC screening.

**Figure 4. Participants’ preferred resources and CPD modalities in general (a), to learn more about risk-based breast cancer (BC) screening (b) and topics to be included in resource material (c) (participants invited to check all that apply)**

**4. Discussion**

This study provides important information on familiarity with the concept of PRS, perceived level of knowledge regarding risk-stratified BC screening, and preferences for CPD of HCPs not trained in genetics. Overall, HCPs reported low level of familiarity with the concept of PRS and limited knowledge regarding risk-stratified BC screening. The vast majority acknowledged their needs for CPD on these topics and would favor resources delivered online.

To our knowledge, only two smaller-size studies (i.e. sample sizes of 105<sup>14</sup> and 120<sup>13</sup>) have reported HCPs’ level of familiarity with the concept of PRS. However, both studies were focused primarily on genetic counselors. Thus, the results of these studies are not comparable with ours since our study population was composed mainly of professionals not trained in genetics. Our study complements the evidence generated by these previous studies by providing the perspectives of a diverse group of HCPs and by highlighting the fact that, unlike genetic counselors, professionals not trained in genetics currently report a low level of familiarity with concept of PRS. Having basic knowledge regarding the calculation and implications of a PRS is important for several HCPs, including front line professionals such as nurses and primary care physicians. Indeed, if calculations of PRS are implemented in clinical practice, they would need to answer questions related to PRS results and support their patients in their

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203 decision process regarding appropriate screening recommendations and preventative options. HCPs  
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1373 vast majority of participants answered that: i) they do not have enough knowledge (60.5%), ii) they would  
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1574 require more training (76.9%), and iii) that the ideal medical and nursing curriculum should include more  
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1775 on this topic (70.3% and 71.3%, respectively) (Figure 3). However, only 45.9% answered that they would  
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should also be knowledgeable of the potential limitations of PRS and be able to convey a balanced message to their patients.<sup>27</sup> Finally, according to different possible implementation scenarios, front line HCPs may have an important role in identifying and referring individuals for whom a risk assessment that included a PRS calculation is most indicated.<sup>15,28</sup>

The observation that the vast majority of our participants stated that they do not have enough knowledge about risk-stratified BC screening and would require more training is in line with several studies reporting that HCPs feel unprepared and lack the appropriate knowledge to competently integrate emerging genomic information into their practice.<sup>14-16,29-32</sup> Scientific literature about the concept of PRS and risk-stratified BC screening has been published since at least 2015.<sup>33,34</sup> Thus, reported low level of familiarity and knowledge of HCPs about these two aspects suggests that active dissemination strategies are required.

To ensure a successful integration of the PRS and risk-stratified screening approaches, a comprehensive portfolio of CPD activities – adapted to the different professional groups and medical specialties – is necessary. Academic institutions will probably need to adapt their curriculum to address these knowledge gaps, and authoritative associations should be called upon to provide point-of-care resources, clinical guidelines, and implementation protocols for the responsible use of PRS information and sound implementation of risk-stratified BC screening.<sup>12</sup>

Although these analyses were exploratory, it is interesting to note significant differences observed on the level of familiarity with the concept of PRS according to participants’ profession, medical specialty, and region of practice. Previous studies assessing level of knowledge with genetics and/or genomics have reported similar differences across professions<sup>30,35,36</sup>, medical specialties<sup>29,37,38</sup> and geographical locations.<sup>37,39,40</sup> These differences on the level of familiarity with the concept of PRS are

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ervices professionals are offering as well as their exposure to genetic services within their health care  
institutions.<sup>36</sup> Knowledgeable of the potential limitations of PRS and has implemented a High Risk  
Message Board for patients.<sup>27</sup> Finally, a thorough review of possible implementation issues for HCPs, front lines  
related to genetics and genomics risk of BC. Uncovering professional group differences on the familiarity  
and knowledge about the concept of PRS and risk- stratified BC screening should be explored further  
through a more comprehensive assessment among different HCPs. This could serve as a parameter of  
guidance for the development of tailored CPD activities and resources.

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Our participants’ preference for online CPD resources is in line with other studies<sup>14,29,41</sup>, although the in-person CPD modality was preferred in one.<sup>42</sup> We may hypothesize that HCPs’ preference for the online modality has probably increased since the occurrence of the Covid-19 pandemic. Online resources have the advantage of being available “just in time”, exactly when HCPs need it and are ready to integrate this in their practice and skill set. This immediacy feature is coherent with an important adult learning theory principle which suggests that learners are interested in acquiring knowledge that ~~have~~has immediate relevance and impact.<sup>43</sup>

#### 4.1. Strengths and Limitations

To our knowledge, the sample size of this study is the largest and includes the most diversified population of HCPs to date to characterize their level of familiarity and perspectives on the concept of PRS and the risk-stratified BC screening approach. The recruitment method was multifaceted and primarily through professional associations and healthcare institutions. Such a recruitment scale is particularly impressive in the context of the Covid-19 pandemic.

Nevertheless, it is important to recognize the limitations of our study. While our questionnaire was designed to be of 15-minute duration in order to accommodate a target population with many competing priorities, a more detailed knowledge status and learning needs collection tool would be required as this is a crucial step in establishing sound CPD curriculum.<sup>44</sup> It is important to recognize that an online recruitment and data collection strategy may result in a greater proportion of participants preferring electronic CPD. Our sample distribution is not representative of the Canadian healthcare professional population. Supplementary material S4 contrasts the demographics of participants with that of the Canadian physicians and nurses workforce. Our sample has a higher proportion of female and of

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professionals from the province of Québec compared to the National statistics . However, the proportion

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2 ed family medicine as their medical specialty is comparable to the national statistics.

f<sup>2</sup>334 Our participants' preference for online CPD resources is in line with other studies<sup>14,29,41</sup>, although  
4 We believe though that our results offer an indication that professionals not trained in genetics  
p<sup>2</sup>535 the in-person CPD modality was preferred in one.<sup>42</sup> We may hypothesize that HCPs' preference for the  
a<sup>2</sup>736 online modality has probably increased since the occurrence of the Covid-19 pandemic. Online resources  
6 stratified BC screening, and that proper CPD should be planned. In some provinces, our recruitment was  
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r more frequently done through a snowball approach within the vast network of our collaborators. This  
t might have resulted in the recruitment of people already interested by and knowledgeable of the concept  
i of PRS and risk-stratified BC screening approach. If this selection bias is present, it would mean that the  
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real level of familiarity with the concept of PRS and knowledge of risk-stratified BC screening of HCPs is even lower than what we observed.

### 5. Conclusion

Current use of PRS testing is at an early stage of integration.<sup>12</sup> While the risk-stratified BC screening based on information such as the PRS is not yet part of any Canadian provinces or territories' public health measures, it is currently undergoing effectiveness-implementation studies in Canada<sup>5</sup>, as well as in the U.S.<sup>6</sup> and in Europe<sup>7</sup>. There is therefore a window of opportunity for professional associations, healthcare institutions and public health or government agencies overseeing screening programs to proactively plan for knowledge dissemination strategies that will effectively support HCPs involved at different contact points in the integration of this emerging genomics strategy. The fact that a lack of knowledge has been identified as the most frequent barrier to the implementation of genetics and genomics in practices justifies the relevance of investing in workforce preparation and CPD activities tailored to healthcare professionals existing knowledge and clinical practice needs.

### Data Availability

Additional data are available from the corresponding author upon request.

### Acknowledgements

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### Author Contributions

HN, JS, AMC; Supervision: HN; Writing-original draft: JL, ACB, CMB, HN; Writing-review & editing: All authors; Conceptualization: All authors; Data curation: JL, ACB, CMB; Formal analysis: JL, ACB, CMB, HN;

### Ethics Declaration

JS, AMC; Methodology: All authors; Project administration: JL, HN; Resources: The study was approved by the Institutional Review Board of: CHU de Québec–Université Laval (2021-5136). Informed consent was obtained from all participants when they opened the survey link.

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Data were collected in a completely anonymous fashion - the information never had identifiers associated with it .

**Conflict of Interest**

The authors declare no conflict of interest. The funders had no role in the design of the study; in the collection, analyses, or interpretation of data; in the writing of the manuscript, or in the decision to publish the results.

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**Figure**

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**Table 1. Participants' characteristics (N=593)**

	<b>Frequency n (%)</b>
<b>Sociodemographic and professional status</b>	
<b>Gender</b>	
<i>Women</i>	432 (93.5)
<i>Men</i>	30 (6.5)
[Missing data/ Prefer not to answer]	[131]
<b>Profession</b>	
<i>Physician</i>	103 (22.3)
<i>Nurse</i>	323 (69.7)
<i>Other<sup>1</sup></i>	37 (8.0)
[Missing data]	[130]
<b>Medical specialty</b>	
<i>Family medicine/Primary care</i>	167 (36.1)
<i>Oncology</i>	59 (12.8)
<i>Other<sup>2</sup></i>	236 (51.1)
[Missing data]	[131]
<b>Number of years of practice</b>	
<i>&lt; 5 years</i>	58 (12.5)
<i>5-14 years</i>	135 (29.2)
<i>15-25 years</i>	113 (24.4)
<i>&gt; 25 years</i>	157 (33.9)
[Missing data]	[130]
<b>Region of practice</b>	
<i>Province of Québec</i>	377 (82.9)
<i>Province of Ontario</i>	46 (10.1)
<i>Other Provinces</i>	32 (7.0)
[Missing data]	[138]
<b>Practice setting</b>	
<i>Academic hospital</i>	133 (28.9)
<i>Community hospital</i>	98 (21.3)
<i>Family health team/group/network</i>	75 (16.3)
<i>Community health centre</i>	78 (17.0)
<i>Private clinic</i>	25 (5.4)
<i>Other<sup>3</sup></i>	51 (11.1)
[Missing data]	[133]
<b>Level of agreement with statement: “Breast cancer screening is an effective method for early detection of breast cancer”</b>	
<i>Agree or strongly agree</i>	528 (89.3)
<i>Neither agree nor disagree</i>	25 (4.2)
<i>Disagree or strongly disagree</i>	31 (5.3)
<i>Don't know</i>	7 (1.2)
[Missing data]	[2]

<sup>1</sup> Other professions include genetic counsellor, physiotherapist, occupational therapist, medical imaging, researcher, and technologist. <sup>2</sup> Other medical specialties includes: internal medicine, surgery, emergency, palliative care, public health medicine, radiology, and obstetrics – gynecology.

<sup>3</sup> Other practice settings include intensive care unit, nurse practitioner-led clinic, nursing home, public health agency, and research center.

## **Supplementary Material S1. Listing of Professional Associations or Organisations who Disseminated the Study Invitation**

Fédération des médecins omnipraticiens du Québec
Canadian Society of Breast Imaging
Association des radiologistes du Québec
CHU de Québec-Université Laval
Ordre des infirmières de recherche
Génome Québec
McGill Faculty of Medicine and Health Sciences Electronic Newsletter
McGill Department of Family Medicine Monthly Newsletter
Infolettre de Pulsar
Ordre des infirmières et infirmiers du Québec
Nurse Practitioners' Association of Ontario
Canadian Partnership Against Cancer
Canadian Association of Medical Oncologists
Canadian Association of Nurses in Oncology
BC College of Family Physicians
Registered Nurses Association of The Northwest Territories and Nunavut
Association of Regulated Nurses of Manitoba
Le collège des médecins de famille de Terre-Neuve-et-Labrador



# PERSPECTIVE

## INTEGRATION & IMPLEMENTATION

### Personalized breast cancer risk assessment and screening mammogram (PERSPECTIVE I & I)

#### Questionnaire for healthcare professionals

If you have any questions regarding this questionnaire, please contact us by email at: [info@etudeperspective.ca](mailto:info@etudeperspective.ca)

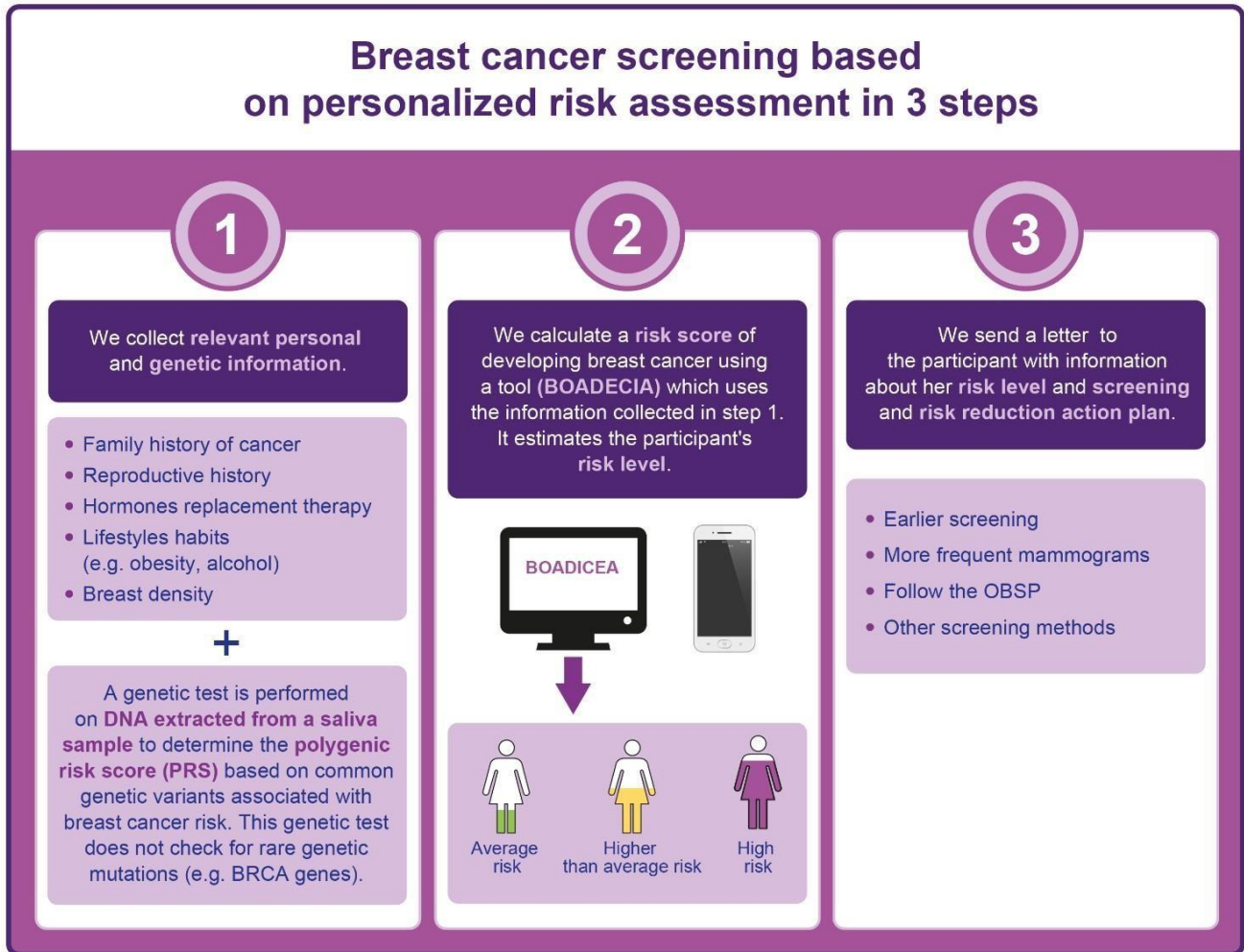
#### About this questionnaire

- Researchers from University of Toronto, Université Laval, McGill University, and University College London have developed this questionnaire. It aims to gather the opinions, attitudes and expectations of healthcare professionals regarding a breast cancer screening approach based on an individual's risk of developing breast cancer in the future. We therefore invite you to answer this short questionnaire, which will take between **12 to 15 minutes**.
- **By completing this questionnaire, you consent to participate in this research.** The information you send will be kept strictly confidential and your identity will remain anonymous. **Please do not include any personal identifiable information (e.g. name).**
- This study is part of a larger project funded by Canadian Institutes for Health Research, Genome Canada, Genome Québec, Ontario Research Fund, the Quebec Breast Cancer Foundation as well as other partners and is aimed at understanding whether there is a benefit to women knowing their breast cancer risk and using this information to make an informed choice about breast cancer screening.
- There are **no right or wrong answers**, and we ask that you simply check off the answers that most apply to you. We encourage you to answer every question. If you come to a question that you do not want to answer, please skip that question and answer the remaining questions.



**Preamble:**

Although screening has benefits such as reducing mortality through early detection, there are also potential harms including overdiagnosis (diagnosis and treatment of breast cancer that would never have been life-threatening). A risk-based breast cancer screening approach is being considered by the scientific community as an option to improve the benefit-harm balance of existing screening programs. This approach would have three stages, as shown in the diagram below:



For more information about the project, visit the project website at: [www.cancercareontario.ca/breastriskstudy](http://www.cancercareontario.ca/breastriskstudy).

**Q1. The Canadian Task Force on Preventive Health Care recommends that women at average risk aged 50-74 be screened with mammography every 2 years. Please, indicate how strongly you agree or disagree that breast screening is an effective method for early detection of breast cancer:**

- Strongly agree
- Agree
- Neither agree nor disagree
- Disagree
- Strongly disagree
- Don't know

**Q2. Have you ever heard of the Personalized Risk Assessment for Prevention and Early Detection of Breast Cancer: Integration and Implementation (PERSPECTIVE I&I) study? (Check all that apply)**

- I have never heard of it before today
- I have heard about it in my clinical practice
- I have heard about it through a website or social media
- I have heard about it through a newsletter
- I have attended a presentation about the study
- I have at least one patient in the study
- Other (please specify): \_\_\_\_\_

**Q3. Breast cancer risk assessment proposed in PERSPECTIVE I&I is based, among other factors, on a polygenic risk score (PRS), measured from a few hundreds of common breast cancer susceptibility genetic variants [single-nucleotide polymorphisms (SNPs)]. How familiar are you with the concept of PRS?**

- Very familiar
- Familiar
- Unfamiliar
- Very unfamiliar
- I don't know this concept

**Q4. Breast cancer screening based on personalized risk assessment aims to adapt screening recommendations as a function of individual risk. Please indicate how strongly you agree or disagree with the following recommendations:**

Recommendations	Strongly agree	Agree	Neither agree or disagree	Disagree	Strongly disagree
For women who are at <b>high risk</b> of breast cancer, <b>increase the frequency</b> of breast screening	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
For women who are at <b>high risk</b> of breast cancer, <b>start the breast screening at an earlier age</b>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
For women who are at <b>higher than average risk</b> of breast cancer, <b>increase the frequency</b> of breast screening	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
For women who are at <b>lower than average risk</b> of breast cancer, <b>decrease the frequency</b> of breast screening	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
For women who are at <b>much lower than average risk</b> of breast cancer, <b>delay the start of breast cancer screening</b>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
For women who are at <b>much lower than average risk</b> of breast cancer, <b>do not offer</b> breast screening	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**Q5a. Women’s risk of developing breast cancer will be estimated using several individual factors such as family history of cancer, breast density, results of a genetic test to measure the polygenic risk score (PRS) and other risk factors. Please read the following statements and indicate whether you think this is a part of your scope of practice (check yes or no):**

Activities	Under the scope of my practice
Discuss the advantages and limitations of personalized breast cancer risk assessment	<input type="checkbox"/> Yes <input type="checkbox"/> No
Collect patient information required to perform a breast cancer risk assessment	<input type="checkbox"/> Yes <input type="checkbox"/> No
Discuss the results of a breast cancer risk assessment with a patient	<input type="checkbox"/> Yes <input type="checkbox"/> No
Explain to your patients the difference between the risk of developing breast cancer in the future and having a diagnosis of breast cancer	<input type="checkbox"/> Yes <input type="checkbox"/> No

**5b. Indicate how comfortable you would be based on your current knowledge to perform these activities with your patients:**

Activities	Very comfortable	Comfortable	Neither comfortable or uncomfortable	Uncomfortable	Very uncomfortable
Discuss the advantages and limitations of personalized breast cancer risk assessment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Collect patient information required to perform a breast cancer risk assessment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Discuss the results of a breast cancer risk assessment with a patient	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Explain to your patients the difference between the risk of developing breast cancer in the future and having a diagnosis of breast cancer	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**Q6. In your opinion, what 3 (three) aspects of the Ontario healthcare system should be enhanced to implement breast cancer screening based on personalized risk assessment? (Check the three most important aspects in your opinion).**

- Number of primary care physicians
- Number of nurse practitioners
- Number of genetic counsellors
- Number of geneticists
- Remuneration of healthcare professionals
- Medical training
- Time allocated to a patient-physician appointment
- Time allocated to a patient-nurse practitioner appointment
- Access to a primary care physician
- Access to a nurse or nurse practitioner
- Access to breast screening (e.g. mammogram, MRI)
- Other (please specify): \_\_\_\_\_
- None, I believe the healthcare system is ready

**Q7. In your opinion, which healthcare professionals should play a role if breast cancer screening based on personalized risk assessment was implemented? (Check all that apply)**

- Primary care physician
- Radiologist
- Surgeon
- Oncologist
- Nurse practitioner
- Geneticist
- Genetic counsellor
- Nurse navigator
- Other (please specify): \_\_\_\_\_

**Q8. Based on your knowledge, what is your opinion regarding breast cancer screening based on personalized risk assessment, including risk calculation and interpretation? Please, indicate how strongly you agree or disagree with the following statements:**

Statements	Strongly agree	Agree	Neither agree, or disagree	Disagree	Strongly disagree
I have enough knowledge regarding personalized breast cancer risk assessment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I require more training on personalized breast cancer risk assessment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I have time to educate myself on personalized breast cancer risk assessment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I think it would be important to include more education on risk assessment, including genetic factors, in the medical curriculum	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I think it would be important to include more education on risk assessment, including genetic factors, in the nursing curriculum	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

**Q9. When you have general questions related to your clinical practice, what resources do you prefer to use? (Check all that apply)**

- Scientific publications
- Training (courses, online conference or in-person)
- Internet search engines
- Colleagues
- Government agencies
- Other (please specify): \_\_\_\_\_

**Q10. For learning more about breast cancer screening based on personalized risk assessment, please select the three resource formats you find most useful for your clinical practice:**

- In-person training such as workshops
- Online courses
- Webinar type conference
- Consultations with a geneticist or a genetic counselor
- An application for your phone or tablet
- Printed material
- Website
- Other (please specify): \_\_\_\_\_

**Q11. Concerning the resources in the previous question, what type of information would you like to find for your clinical practice? (Check all that apply)**

- General information on genetics
- Information on common genetic variants (SNPs)
- Information on the basics of personalized breast cancer risk assessment
- Information on the calculation of a polygenic risk score (PRS)
- Information on interpreting results of breast cancer risk assessment
- Information on the best practices of breast cancer risk level communication
- Information on breast cancer prevention
- Information on the main ethical, legal and social challenges of personalized breast cancer risk assessment
- Other (please specify): \_\_\_\_\_

**Q12. What is your gender?**

- Female
- Male
- Other
- Prefer not to answer

**Q13. What is your profession?**

- Physician
- Nurse practitioner
- Nurse navigator
- Genetic Counsellor
- Other (please specify): \_\_\_\_\_

**Q14. What is your main medical specialty?**

- Family medicine/ Primary care
- Geriatrics
- Genetics
- Internal medicine
- Public health medicine
- Surgery
- Radiology
- Medical oncology
- Surgical oncology
- Obstetrics - Gynecology
- Gynecologic Oncology
- Other (please specify): \_\_\_\_\_

**Q15. For how long have you been practicing your profession?**

- Less than 5 years
- Between 5 and 9 years
- Between 10 and 14 years
- Between 15 and 19 years
- Between 20 and 25 years
- More than 25 years

**Q16. What is your main institution of practice?**

- Academic hospital
- Community hospital
- Family health team (Family Health Organizations (FHOs), Family Health Networks (FHNs))
- Family Health Group (FHGs)
- Community Health Centre (CHCs)
- Comprehensive Care Models (CCMs)
- Nurse practitioner-led clinic (NPLCs)
- Private clinic
- Other (please specify): \_\_\_\_\_

**Q17. In which Provinces or Territories do you mainly practice?**

- Ontario
- Québec
- British Columbia
- Alberta
- Manitoba
- Saskatchewan
- Prince Edward Island



- New Brunswick
- Nova Scotia
- Newfoundland and Labrador
- Northwest Territories
- Yukon
- Nunavut

**Do you have any comments or suggestions?**

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**Thank you for your participation!**

**Supplemental Material S3. Association between familiarity with PRS and sociodemographic variables (n = 593)**

<b>Variables</b>	<b>Very familiar or familiar n = 46, (%)</b>	<b>Very unfamiliar or unfamiliar n = 307, (%)</b>	<b>Don't know this concept n = 240, (%)</b>	<b>p-value</b>
<b>Gender</b>				0.05
<i>Women</i>	31 (7.2)	223 (51.6)	178 (41.2)	
<i>Men</i>	5 (16.7)	20 (66.7)	5 (16.7)	
<i>Prefer not to answer/Missing</i>	10 (7.6)	64 (48.9)	57 (43.5)	
<b>Profession</b>				<.01
<i>Physician</i>	19 (18.4)	60 (58.3)	24 (23.3)	
<i>Nurse</i>	13 (4.0)	157 (48.6)	153 (47.4)	
<i>Other</i>	4 (10.8)	27 (73.0)	6 (16.2)	
<i>Missing</i>	10 (7.7)	63 (48.5)	57 (43.8)	
<b>Professional specialty</b>				0.02
<i>Family medicine/Primary care</i>	10 (6.0)	85 (50.9)	72 (43.1)	
<i>Oncology</i>	12 (20.3)	29 (49.2)	18 (30.5)	
<i>Other</i>	14 (5.9)	130 (55.1)	92 (39.0)	
<i>Missing</i>	10 (7.6)	63 (48.1)	58 (44.3)	
<b>Number of years of practice</b>				0.18
<i>&lt; 5 years</i>	9 (15.5)	32 (55.2)	17 (29.3)	
<i>5-14 years</i>	5 (3.7)	77 (57.0)	53 (39.3)	
<i>15-25 years</i>	10 (8.8)	54 (47.8)	49 (43.4)	
<i>&gt; 25 years</i>	12 (7.6)	81 (51.6)	64 (40.8)	
<i>Missing</i>	10 (7.7)	63 (48.5)	57 (43.8)	
<b>Region of practice</b>				0.04
<i>Province of Québec</i>	23 (6.1)	191 (50.7)	163 (43.2)	
<i>Province of Ontario</i>	7 (15.2)	26 (56.5)	13 (28.3)	
<i>Other Provinces</i>	5 (15.6)	20 (62.5)	7 (21.9)	
<i>Missing</i>	11 (8.0)	70 (50.7)	57 (41.3)	
<b>Practice setting</b>				0.13
<i>Academic hospital</i>	17 (12.8)	71 (53.4)	45 (33.8)	
<i>Community hospital</i>	6 (6.1)	54 (55.1)	38 (38.8)	
<i>Family health team/group/network</i>	3 (4.0)	45 (60.0)	27 (36.0)	
<i>Community health centre</i>	4 (5.1)	33 (42.3)	41 (52.6)	
<i>Private clinic</i>	2 (8.0)	15 (60.0)	8 (32.0)	
<i>Other</i>	4 (7.8)	24 (47.1)	23 (45.1)	
<i>Missing</i>	10 (7.5)	65 (48.9)	58 (43.6)	

**Table S4. Comparison between participating physicians and nurses and Canadian workforce 2020 data<sup>1</sup>**

	<b>Physicians</b>		<b>Nurses<sup>2</sup></b>	
	Study sample n=103	Canadian workforce N=92,173	Study sample n=323	Canadian workforce N=448,044
	n (%)	n (%)	n (%)	n (%)
<b>Sociodemographic and professional status</b>				
<b>Gender</b>				
<i>Women</i>	80 (78.4)	40,280 (43.7)	316 (98.1)	392,039 (87.5)
<i>Men</i>	22 (21.6)	51,893 (56.3)	6 (1.9)	56,001 (12.5)
[Missing data/ Prefer not to answer]	[1]	[0]	[1]	[0]
<b>Medical specialty<sup>3</sup></b>				
<i>Family medicine</i>	56 (57.3)	46,797 (50.8)	109 (37.6)	Not available
<i>Other specialty</i>	47 (42.7)	45,376 (49.2)	181 (62.4)	Not available
[Missing data]	[0]	[0]	[33]	
<b>Region of practice</b>				
<i>Province of Québec</i>	76 (76.8)	22,038 (23.9)	276 (85.7)	103,421 (23.1)
<i>Province of Ontario</i>	15 (15.1)	33,830 (36.7)	27 (8.4)	162,760 (36.3)
<i>Other Provinces</i>	8 (8.1)	36,305 (36.4)	19 (5.9)	181,863 (40.6)
[Missing data]	[4]	[0]	[1]	[0]

<sup>1</sup> Data obtained from the Canadian Institute for Health Information. Supply, Distribution and Migration of Physicians in Canada, 2020 — Data Tables. Ottawa, ON: CIHI; 2021 and the Canadian Institute for Health Information. Nursing in Canada, 2020 — Data Tables. Ottawa, ON: CIHI; 2021.

<sup>2</sup> In Canada, regulated nursing professionals comprise four groups, namely nurse practitioners, registered nurses, registered psychiatric nurses and licensed practical

nurses.

<sup>3</sup>For nurses, medical specialty data were available for registered nurses only. This group represents 304,558 individuals.