Genetic testing practices among specialist physicians who treat prostate cancer: Updated survey of Canadian practitioners

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KEY MESSAGES

- In a survey of academic, multidisciplinary specialist physicians, 90% offered genetic testing for PCa, representing an increase in awareness, knowledge, and access from the initial survey in 2022.
- Testing rates have increased across all PCa disease states, suggesting a growing consensus that broader testing of patients beyond current guideline recommendations may be clinically valuable.
- The most common forms of genetic testing were germline and somatic testing, although an increase in the rates of ctDNA testing reflects its growing implementation in clinical practice.
- Despite improved access through provincial funding, gaps remain, with nearly 20% of physicians still relying on private pay or clinical trials to obtain genetic testing.

INTRODUCTION

Genetic testing has become important in prostate cancer (PCa) management. Globally, there are disparities and barriers to implementation of testing.[1-3] Current Canadian guidelines recommend genetic testing for high-risk patients and patients where results may inform treatment.[4]

A 2022 survey highlighted the growth of genetic testing and described testing practices and access among Canadian physicians treating PCa.[5] Given the evolving treatment landscape of PCa, a follow-up survey was conducted to identify changes in PCa genetic testing practices over time.

METHODS

The Canadian Genitourinary Research Consortium (GURC) developed a cross-sectional survey to assess the PCa genomic testing landscape in Canada. Principal investigators (PIs) and sub-investigators (sub-Is) of the GURC study were invited to participate.[6] The initial survey was conducted January to June 2022, and the refresh was conducted July to September 2023.The survey included 21 questions to capture changes in genomic testing, as described previously.[5] Patient-level data were not collected, and data collection from study investigators via survey was approved by Research Ethics Boards at GURC sites. Analyses were descriptive; the number (proportion) of responses in each category were reported.

RESULTS

A total of 30 investigators from 22 sites responded, including 21/22 PIs (95% response rate) and 9 sub-Is, with 12 urologic oncologists, 14 medical oncologists and two radiation oncologists from six provinces, with most (87%) practicing in academic settings.

Current practices in genetic testing for prostate cancer

Among respondents who offered genetic testing (90%), germline and somatic testing were most common, offered by >60% of physicians (Table 1). Most physicians (53%) initiated genetic testing themselves versus referring to a specialist. Results of testing impacted patient management for all physicians who offered testing. Most physicians (63-85%) referred patients with an alteration of significance to a genetics specialist. Biobanking was uncommon; less than one-third of physicians banked samples regardless of genetic variant.

Physicians were asked about patients receiving genetic testing; 85% reported that >40% of patients with metastatic castration-resistant PCa (mCRPC) received testing, followed by 67% for metastatic castration-sensitive PCa (mCSPC), 26% for non-mCRPC (nmCRPC), 15% for high-risk PCa, and 7% for any PCa.

Recommended practices

All physicians agreed that genetic testing should be offered in PCa, dependent on patient and disease characteristics; the most ideal tests were ctDNA/cfDNA (89%), germline (85%) and tumour DNA damage repair (85%) (Table 2). Most (73%) agreed that academic medical oncologists should offer testing for patients, although consensus was not reached around which other specialties should offer testing.

Patient population recommended for genetic testing

Genetic testing was recommended across all disease states by at least some physicians (Table 3). Although responses were mixed for the most appropriate tests, most physicians (50-63%) agreed that mCSPC and mCRPC patients should be offered ctDNA, germline, and somatic testing. Most respondents (57-60%) would offer germline testing for localized PCa if the patient had a primary relative with PCa, ovarian, breast or pancreatic cancer; over one-third (36-40%) would do so for a patient with any relative with those cancers.

Timing of genetic testing

While <10% of physicians recommended testing at PCa diagnosis, this proportion increased up to 41% for patients with a family history of prostate, breast, ovarian, or pancreatic cancer (Table 4). At nmCRPC diagnosis, 37% of respondents recommended germline and 30% recommended tumour or ctDNA testing. At mCSPC or mCRPC diagnosis, 67-70% recommended germline testing and 89% recommended tumour or ctDNA testing.

DISCUSSION

This follow-up survey of 30 multidisciplinary specialist physicians primarily from academic centres reveals a continued evolution of the PCa genetic testing landscape in Canada. Compared with the 2022 survey, this 2023 follow-up highlights increased awareness, knowledge, and access to testing, which has translated to greater initiation of mainstream germline and somatic testing and more frequent ctDNA testing.

In this updated survey, 90% of respondents offered genetic testing for PCa, an increase from 84% in 2022. While germline and somatic remained the most available forms of testing, more clinicians offered ctDNA testing (26%, up from 0%), reflecting its growing implementation in clinical practice.[7] More physicians initiated genetic testing themselves (53%, up from 34%) versus referring to a genetic specialist, potentially reducing barriers to access. While clinicians recognized the importance of testing, 70% would not offer genetic testing to family members of patients with a genomic alteration, and 90% rely on genetic counsellors to fill this gap. Only 26% of physicians would offer germline testing for a patient with a familial history of PCa, suggesting that clinicians understand the difference between familial and germline genetic associations. Compared with the previous survey, more physicians referred patients to a genetics specialist when significant genetic alterations were detected in genes such as CDK12, CHEK2, and PALB2, and for variants of uncertain significance, although referral practices for less-characterized alterations remains uncertain.

The 2023 Canadian guidelines for genetic testing recommend genetic testing for patients with metastatic disease, high-risk localized disease, or relevant family history.[4] In concordance with recommendations, more physicians reported that >40% of their patients received testing for mCRPC (85%, up from 52%), mCSPC (67%, up from 42%), nmCRPC (26%, up from 12%), and high-risk PCa (15%, up from 3%) compared with 2022. This shift could suggest a growing consensus that broader testing beyond current guidelines may be valuable.

Physicians reported increased provincial funding for genetic testing, with 70% reporting public funding access, up from 63% in 2022, corresponding to decreased reliance on clinical trials (19%, down from 63%) and private pay (19%, down from 31%). However, nearly 1/5 physicians still rely on these alternative funding avenues, indicating that access gaps remain and current provincial funding does not fully meet demands.

Limitations of this study include its descriptive nature and that it did not capture all Canadian regions. Future investigation into regional discrepancies and barriers to testing is warranted.

This follow-up survey demonstrates progress in the integration of genetic testing into PCa care in Canada. Physicians reported greater access to and knowledge in all types of genetic testing, and growing consensus around expanding testing across the disease spectrum. Despite advances, there remains opportunities for physician education, particularly in access, funding, results interpretation and referral pathways.

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FIGURES AND TABLES

Table 1. Current practices for the genomic testing of patients with prostate cancer		
Survey question	**2023 survey n (%)**	**2022 survey[5] n (%)**
Province (n=30)		
Alberta	7 (23%)	8 (21%)
British Columbia	9 (30%)	11 (29%)
Manitoba	2 (7%)	5 (13%)
Nova Scotia	1 (3%)	1 (3%)
Ontario	8 (27%)	8 (21%)
Quebec	3 (10%)	5 (13%)
Is mainstream genetic testing initiated by clinicians or is a referral necessary? (n=30)		
Initiated by clinicians	16 (53%)	13 (34%)
Referral to a specialist	10 (33%)	17 (45%)
Other	4 (13%)	8 (21%)
Do you offer genetic testing to family members of patients with a positive genomic alteration? (n=27)		
Yes	8 (30%)	8 (24%)
No	19 (70%)	24 (73%)
If no, who offers this? (n=19)		
Genetics	17 (90%)	17 (71%)
Other	2 (11%)	N/A
What form of genetic testing is currently available to you?* (n=27)		
Germline	24 (89%)	30 (91%)
Tumor DNA damage repair genetic testing	25 (93%)	19 (58%)
IHC Staining	6 (22%)	4 (12%)
ISH	4 (15%)	0 (0%)
ctDNA/cfDNA	9 (33%)	10 (30%)
Other	1 (4%)	3 (9%)
I do not know	0 (0%)	
Do you currently perform/offer genetic testing in your PCa patients? (n=30)		
Yes	27 (90%)	32 (84%)
No	3 (10%)	6 (16%)
If yes, which type of testing do you perform/offer?* (n=27)		

Germline DNA damage repair genetic testing	17 (63%)	30 (94%)
Tumor DNA damage repair genetic testing	18 (67%)	23 (72%)
ctDNA DNA damage repair genetic testing	7 (26%)	N/A
Other	3 (11%)	1 (3%)
If yes, how do you arrange for testing?* (n=27)		
Clinical trials	6 (22%)	20 (63%)
Private pay	5 (19%)	10 (31%)
Provincially funded testing	19 (70%)	20 (63%)
Other	1 (4%)	9 (28%)
Do results of genomic testing impact your patient management? (n=27)		
Yes	27 (100%)	32 (97%)
No	0 (0%)	1 (3%)
If yes, it will change my patient's treatment plan and sequencing of the following: (n=27)		
Chemotherapy	3 (11%)	2 (6%)
PARP inhibitor therapy	9 (33%)	9 (28%)
Other	11 (41%)	14 (44%)
Missing	4 (15%)	4 (13%)
Do you refer to a genetics specialist for *BRCA1/2* or *ATM*? (n=27)		
Yes	26 (96.3%)	31 (94%)
No	1 (3.7%)	2 (6%)
Do you refer to a genetics specialist for BRIP1, BARD1, CDK12, CHEK1, CHEK2, FANCL, PALB2, PPP2R2A, RAD51B, RAD51C? (n=27)		
Yes	23 (85%)	21 (64%)
No	4 (15%)	10 (30%)
Do you refer when any other genetic alteration is identified? (n=27)		
Yes	17 (63%)	13 (39%)
No	10 (37%)	19 (58%)
Do you refer when any variants of unknown significant (VUS) are identified? (n=27)		
Yes	11 (41%)	9 (27%)
No	16 (59%)	23 (70%)
Do you biobank or maintain a database/registry when the following are identified? (n=27)		

BRCA/ATM		
Yes, biobank	8 (30%)	12 (36%)
Yes, database/registry	8 (30%)	9 (27%)
No	10 (37%)	11 (33%)
Missing	1 (4%)	1 (3%)
BRIP1, BARD1, CDK12, CHEK1, CHEK2, FANCL, PALB2, PPP2R2A, RAD51B, RAD51C, RAD51D, RAD54L alteration		
Yes, biobank	7 (26%)	12 (36%)
Yes, database/registry	5 (19%)	6 (18%)
No	14 (52%)	14 (42%)
Missing	1 (4%)	1 (3%)
VUS		
Yes, biobank	4 (15%)	9 (27%)
Yes, database/registry	1 (4%)	6 (18%)
No	20 (74%)	16 (49%)
Missing	2 (7%)	2 (6%)

Note: n reported for each question reflects responses for the 2023 survey. Details on responses for the 2022 survey have been previously reported.[5] N/A indicates a response that was not included in the 2022 survey. [*]Investigators were allowed to select more than one option. [**]Example, HOXB. *ATM*: ataxia telangiectasia mutated; *BARD1*: BRCA associated RING domain 1; *BRCA1/2*: breast cancer type1/2 susceptibility gene; *BRIP1*: *BRCA1* interacting protein C-terminal helicase 1; *CDK12*: cyclin-dependent kinase inhibitor 12; *CHEK:* checkpoint kinase; ctDNA/cfDNA: circulating tumor/cell free DNA; *FANCL*: Fanconi anemia, complementation group L; IHC: immunohistochemistry; ISH: in-situ hybridization; PALB2: partner and localizer of *BRCA2*; PARP: poly (ADP-ribose) polymerase; PCa: prostate cancer; PPP2R2A: serine/threonine-protein phosphatase 2A regulatory subunit B55α; VUS: variants of unknown significant.

Table 2. Recommended practices for genetic testing of patients with prostate cancer		
Survey question	**2023 survey n (%)**	**2022 survey[5] n (%)**
In your opinion, should genetic testing be offered to PCa patients in your practice?* (n=30)		
Yes, ideally germline (inherited) DNA damage repair genetic testing alone	12 (40%)	3 (8%)
Yes, ideally tumor tissue DNA damage repair genetic testing alone	11 (37%)	N/A
Yes, ideally circulating tumor DNA for DNA damage repair testing	15 (50%)	N/A
In your opinion, what is the ideal form of genetic testing that should be offered?* (n=27)		
Germline	23 (85%)	26 (79%)
Tumour DNA damage repair genetic testing	23 (85%)	25 (76%)
IHC Staining	4 (15%)	3 (9%)
ISH	5 (19%)	2 (6%)
ctDNA/cfDNA	24 (89%)	17 (52%)
Other	1 (4%)	1 (3%)
I don't know	1 (4%)	2 (6%)
Should combination genetic testing be performed? (n=27)		
Yes, ctDNA, followed by or supplemented by tumor tissue testing	7 (26%)	14 (42%)
Yes, tumour tissue testing followed by or supplemented by ctDNA biopsy	14 (52%)	8 (24%)
Yes, but only in some circumstances	2 (7%)	1 (3%)
I don't know	2 (7%)	8 (24%)
No	1 (4%)	N/A
Missing	1 (4%)	2 (6%)
What specialties do you feel should offer genetic testing for patients diagnosed with PCa?* (n=30)		
Non-academic urologist	11 (37%)	9 (24%)
Non-academic medical oncologist	16 (53%)	13 (34%)
Non-academic radiation oncologist	13 (43%)	7 (18%)
Academic urologist	18 (60%)	17 (45%)
Academic medical oncologist	22 (73%)	20 (53%)
Academic radiation oncologist	15 (50%)	14 (37%)

All clinicians who treat PCa	12 (40%)	18 (47%)
All clinicians who treat PCa who have training with genetic testing	16 (53%)	14 (37%)
Genetics specialists	14 (47%)	14 (37%)

Note: n reported for each question reflects responses for the 2023 survey. Details on responses for the 2022 survey have been previously reported.[5] N/A indicates a response that was not included in the 2022 survey. *Investigators were allowed to select more than one option. ctDNA/cfDNA: circulating tumor/cell free DNA; ISH: in-situ hybridization; PCa: prostate cancer

Table 3. Profile of patients who should ideally be offered genetic testing								
Patient characteristics*	**2023 survey (n = 30)**				**2022 survey[5] (n=38)**			
	LPCa n (%)	**mCSPC n (%)**	**mCRPC n (%)**	**nmCRPC n (%)**	**LPCa n (%)**	**mCSPC n (%)**	**mCRPC n (%)**	**nmCRPC n (%)**
All PCa patients								
ctDNA	1 (3%)	19 (63%)	19 (63%)	12 (40%)	N/A	N/A	N/A	N/A
Germline	5 (17%)	15 (50%)	17 (57%)	15 (50%)	3 (8%)	1 (3%)	1 (3%)	3 (8%)
Somatic	3 (10%)	17 (57%)	18 (60%)	15 (50%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)
High-risk PCa patients only**								
ctDNA	6 (20%)	–	–	9 (30%)	N/A	N/A	N/A	N/A
Germline	11 (37%)	–	–	8 (27%)	5 (13%)	–	–	0 (0%)
Somatic	5 (17%)	–	–	8 (27%)	0 (0%)	–	–	0 (0%)
Patients with a primary relative*** with PCa								
ctDNA	4 (13%)	15 (50%)	14 (47%)	11 (37%)	N/A	N/A	N/A	N/A
Germline	17 (57%)	12 (40%)	13 (43%)	14 (47%)	14 (37%)	2 (5%)	4 (11%)	3 (8%)
Somatic	6 (20%)	11 (37%)	11 (37%)	11 (37%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)
Patients with any relative with PCa								
ctDNA	1 (3%)	11 (37%)	11 (37%)	9 (30%)	N/A	N/A	N/A	N/A
Germline	13 (43%)	10 (33%)	10 (33%)	11 (37%)	5 (13%)	3 (8%)	4 (11%)	3 (8%)
Somatic	3 (10%)	9 (30%)	9 (30%)	10 (33%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)

Patients with a primary relative with ovarian, breast or pancreatic cancer								
ctDNA	3 (10%)	15 (50%)	15 (50%)	12 (40%)	N/A	N/A	N/A	N/A
Germline	18 (60%)	12 (40%)	13 (43%)	13 (43%)	13 (34%)	4 (11%)	5 (13%)	3 (8%)
Somatic	5 (17%)	11 (37%)	11 (37%)	12 (40%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)
Patients with any relative with ovarian, breast or pancreatic cancer								
ctDNA	1 (3%)	12 (40%)	12 (40%)	8 (27%)	N/A	N/A	N/A	N/A
Germline	11 (37%)	10 (33%)	11 (37%)	11 (37%)	6 (15%)	4 (11%)	4 (11%)	3 (8%)
Somatic	3 (10%)	9 (30%)	10 (33%)	9 (30%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)
Patients with a family history of another cancer								
ctDNA	0 (0%)	12 (40%)	12 (40%)	8 (27%)	N/A	N/A	N/A	N/A
Germline	3 (10%)	8 (27%)	7 (23%)	8 (27%)	4 (11%)	3 (8%)	4 (11%)	3 (8%)
Somatic	1 (3%)	7 (23%)	6 (20%)	7 (23%)	0 (0%)	0 (0%)	0 (0%)	0 (0%)

Note: N/A indicates a response that was not included in the 2022 survey. *Investigators were allowed to select more than one option. **High-risk nmCRPC: prostate cancer antigen (PSA) doubling time ≤ 10 months. ***Primary relative: first degree relative.
LPCa: localized prostate cancer; mCRPC: metastatic castration-resistant prostate cancer; mCSPC: metastatic castration-sensitive prostate cancer; nmCRPC: non-metastatic castration-resistant prostate cancer; PCa: prostate cancer.

Table 4. Timing of genetic testing				
Timing of genetic testing*	**2023 survey (n = 27)**		**2022 survey[5] (n = 33)**	
	Germline n (%)	**Tumour or ctDNA n (%)**	**Germline n (%)**	**Tumour or ctDNA n (%)**
PCa diagnosis	2 (7%)	1 (4%)	5 (15%)	2 (6%)
PCa diagnosis for patients with a family history of PCa	7 (26%)	1 (4%)	10 (30%)	3 (9%)
PCa diagnosis for patients with a primary relative with breast, ovarian, prostate or pancreatic cancer	11 (41%)	1 (4%)	16 (49%)	5 (15%)
PCa diagnosis for patients with any relative with breast, ovarian, prostate or pancreatic cancer	8 (30%)	3 (11%)	10 (30%)	5 (15%)
High-risk localized PCa** patients	7 (26%)	4 (15%)	11 (33%)	6 (18%)
All nmCRPC diagnosis	10 (37%)	8 (30%)	12 (36%)	7 (21%)
All nmCRPC diagnosis at PSA progression	9 (33%)	9 (33%)	11 (33%)	6 (18%)
All nmCRPC diagnosis at progression to mCRPC	12 (44%)	12 (44%)	13 (39%)	9 (27%)
All nmCRPC patients with a primary relative with breast, ovarian, prostate or pancreatic cancer	11 (41%)	7 (26%)	12 (36%)	6 (18%)
All nmCRPC patients with any relative with breast, ovarian, prostate or pancreatic cancer	12 (44%)	9 (33%)	13 (39%)	7 (21%)
High-risk nmCRPC*** diagnosis	9 (33%)	8 (30%)	12 (36%)	7 (21%)
High-risk nmCRPC*** diagnosis at PSA progression	8 (30%)	7 (26%)	10 (30%)	6 (18%)
High-risk nmCRPC*** diagnosis at progression to mCRPC	9 (33%)	11 (41%)	11 (33%)	9 (27%)
High-risk nmCRPC*** patients with a primary relative with breast, ovarian, prostate or pancreatic cancer	12 (44%)	9 (33%)	12 (36%)	8 (24%)
High-risk nmCRPC*** patients with any relative with breast, ovarian, prostate or pancreatic cancer	10 (37%)	7 (26%)	11 (33%)	8 (24%)

Table 4. Timing of genetic testing				
Timing of genetic testing*	**2023 survey (n = 27)**		**2022 survey[5] (n = 33)**	
	Germline n (%)	**Tumour or ctDNA n (%)**	**Germline n (%)**	**Tumour or ctDNA n (%)**
mCSPC diagnosis	19 (70%)	24 (89%)	21 (64%)	16 (49%)
mCSPC at progression to mCRPC	16 (59%)	19 (70%)	17 (52%)	18 (55%)
mCSPC patients with a primary relative with breast, ovarian, prostate or pancreatic cancer	17 (63%)	16 (59%)	19 (58%)	11 (33%)
mCSPC patients with any relative with breast, ovarian, prostate or pancreatic cancer	15 (56%)	17 (63%)	15 (46%)	10 (30%)
mCRPC diagnosis	18 (67%)	24 (89%)	23 (70%)	18 (55%)
mCRPC progression on 1st line treatment	16 (59%)	21 (78%)	18 (55%)	19 (58%)
mCRPC progression on 2nd line treatment	16 (59%)	20 (74%)	18 (55%)	18 (55%)
mCRPC patients with a primary relative with breast, ovarian, prostate or pancreatic cancer	17 (63%)	17 (63%)	19 (58%)	13 (39%)
mCRPC patients with any relative with breast, ovarian, prostate or pancreatic cancer	16 (59%)	18 (67%)	15 (46%)	10 (30%)
Patient's age at diagnosis will impact my decision	1 (4%)	3 (11%)	6 (18%)	3 (9%)
Other	1 (4%)	1 (4%)	3 (9%)	1 (3%)

*Investigators were allowed to select more than one option.
**High-risk localized PCa: T3 or higher staging (grade group 4 or 5 – Gleason score 8–10) Lymph Node involvement, PSA ≥ 20.
***High-risk nmCRPC: PSA doubling time ≤10 months.
mCRPC: metastatic castration-resistant prostate cancer; mCSPC: metastatic castration-sensitive prostate cancer; nmCRPC: non-metastatic castration-resistant prostate cancer; PCa: prostate cancer; PSA: prostate-specific antigen.