
Genetic counseling and oncology: proposed approaches for collaborative care delivery

Jacquelyn Powers, MS, Kelsey Spielman, MS, Rebecca Mueller, MS, Melissa Batson, BS, Stacy Pundock, BS, Anna Arutyunova, BS, Heather Symecko, MPH, Susan Domchek, MD

University of Pennsylvania, Bassett Center for BRCA, Philadelphia, Pennsylvania, USA

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Demand for cancer genetic counseling has grown rapidly in recent years as germline genomic information has integrated into cancer care. There are currently an

insufficient number of genetic counselors (GC) to address genetic testing need through traditional pre- and post-test counseling. Alternative genetic counseling frameworks, discussed here, are under study to increase access to genetic testing while optimizing the skillsets of existent master's-trained GCs.

Key Words: genetic counseling, genetic testing, optimization, alternative delivery model

Demand for cancer genetic counseling has grown rapidly in recent years as germline genomic information has integrated into cancer care.¹ There are particular cohorts in which a missed opportunity for genetic testing is a missed opportunity for the potential of a targeted therapeutic intervention.² In particular, this is applicable to men with metastatic prostate cancer with germline *BRCA1* or *BRCA2* mutations as it relates to poly ADP ribose polymerase (PARP) inhibitors, or germline mismatch repair gene mutations as it relates to PD-1 inhibitors. The National Comprehensive Cancer Network (NCCN) has adapted genetic testing guidelines to support the recommendation to extend genetic testing to all men with metastatic prostate cancer regardless of family history^{3,4} and all those with regional disease.³

The traditional approach to the identification of individuals with genetic cancer susceptibility has been risk assessment and genetic testing under the provision of a specialist such as a genetic counselor

(GC). This involves a pre- and post-test consultation in which the patient initially presents to a clinical genetics clinic for review of personal/family history, formulation of a differential diagnosis, facilitation of informed consent and specimen collection. The patient then returns to the clinic for results interpretation and medical management discussion; the latter often coupled with a physician. There are recognized benefits to the traditional model including improved patient satisfaction, adherence to cancer risk management, as well as documented cost savings for an institution.¹ Furthermore, misinterpretation of test results, inappropriate medical management, and adverse psychosocial outcomes have been reported in the absence of adequate genetic counseling.⁵⁻⁷ This traditional framework is considered the standard of care by certain professional organizations;¹ however, in oncological care, this framework is presently challenged by the growing need for genetic testing for therapeutic decision making and the limited GC workforce. This increased need has forced dialogue and the development of strategies for alternative approaches to genetic counseling (e.g. telegenetics, telephone counseling). The latter strategies designed to engage patients and increase GC access, while trying to optimize the skillsets of existent master's-trained GCs.

Address correspondence to Jacquelyn Powers, 3400 Civic Center Blvd., 3W Perelman Center for Advanced Medicine (PCAM), Philadelphia, PA 19104 USA

TABLE 1. Alternative genetic counseling delivery models

	Traditional	Oncology only	Hybrid 1 ⁹	Hybrid 2 Penn/MSKCC ⁸	Other considerations
Pre-test ^a	GC ^d	O ^d	O	Web-based video https://youtube.com/watch?v=80X70KUArRg in collaboration with O IC ^d , sample collection, test order ^e : RS ^d	Training of staff via on-site GC and/or GC consultant Standardized materials with ability for modification as information changes
Post-test ^b	GC +/- MD ^d	O	GC + O	GC +/- MD	Utilization of testing laboratory GC
Follow up ^c	GC	O	GC + O	GC	Automated approaches after review of benefits, risks, limitations (preferred: IRB protocol)

^a**pre-test elements:** Gene-specific information (risk & tumor spectrum; well- or ill-defined), results implications and possibility of uncertainty, implications and inheritance for at-risk family members, insurance and fees, psychosocial assessment, knowledge regarding optimal diagnostic genetic testing laboratories & test selection*, GINA, medical management options, expressed importance of sharing information with at-risk family members, plan for disclosure, signed informed consent.

^b**post-test elements:** Disclosure, interpretation for patient (recommendations for prostate cancer treatment), interpretation for family members, provision of additional cancer screening recommendations.

^c**follow up:** Cascade testing, updates regarding variant reclassification, updates as clinical genetics evolves and expands.

^dGC = genetic counselor; MD = medical doctor; O = Oncologist; IC = informed consent; RS = research staff member

^e**Customized prostate panel consideration:**

- (i) Therapeutic: *ATM, BRCA1, BRCA2, MLH1, MSH2, MSH6, PMS2, EPCAM*
- (ii) Additional: *BRIP1, CHEK2, HOXB13, RAD51C, RAD51D, TP53*

Several centers have proposed different models of genetic evaluation of men with prostate cancer, Table 1. Some institutions have considered weighted involvement of the treating oncologist in pre-test education and GC involvement weighted toward post-test responsibilities. In order for such an approach to be successful providers should have proficiencies with the pre-test elements as listed in the subtext of Table 1 and implementation procedures must be in place in order to integrate into clinic practice flow. The challenges of insurance coverage and test costs, discerning optimal diagnostic testing laboratory (ies), variant reclassification and communication must be considered and balanced realistically with provider bandwidth. An additional hybrid approach is being run in parallel at two academic medical centers; the University of Pennsylvania (Penn) and Memorial Sloan Kettering Cancer Center (MSKCC). Patients with metastatic prostate cancer, through an IRB protocol, receive standardized pre-test education using a video and brochure by non-genetics provider. The primary endpoint is to evaluate the acceptability of an alternative care model, as measured by emotional distress and satisfaction with genetic testing decision and with genetic counseling (analysis underway). The

research staff (RS) facilitates informed consent and biospecimen collection. Per protocol, the genetic test is preselected (14 gene panel) and testing is currently covered by the study. Penn and MSKCC GCs return results and provide post-test telephone counseling. This point of care testing yielded a nearly 9-fold increase in patients who underwent genetic testing in 2018 as compared to pre-protocol "traditional model" usual care in 2017, as well as a 7-fold increase in pathogenic/likely pathogenic variants identified.⁸ As this protocol continues, there will be modifications to address feasibility and sustainability such as cost responsibility and implementation without RS participation, while still relying on GCs for results interpretation, disclosure, and cascade testing, if applicable. Of note, in the absence of on-site GCs, most CLIA-CAP commercial diagnostic laboratories employ their own GCs to whom patients/providers may request telephone genetic counseling. Efficacy data is currently lacking when comparing the traditional versus alternative approaches.

Lastly, and in brief, automated tools are under development to scale delivery of genetic and genomic information. One example is HIPAA-compliant,

clinical grade chatbots (such as www.cleargenetics.com). Through inclusion and review from experts within the key medical communities, including GCs, there is potential for responsible delivery of automated approaches under well-defined clinical scenarios. Prior to standard clinical use, such approaches are best studied under IRB protocol. What remains consistent in clinical cancer genetics is the ever-changing landscape. Continued dialogue across oncology, urology, genetic counseling, as well as commercial laboratories, and direct-to-consumer companies is critically important to address the needs of men undergoing germline testing for inherited PrCa.⁹

Disclosures

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