Recent insights on genetic testing in primary prostate cancer diagnosis

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Supplementary Table 1: Literature overview

Specific sorting of all search results (N=111):

1. Included clinical trial (N=34)

Title	Authors	Journal	Year
A cohort analysis of men with a family history of BRCA1/2 and Lynch mutations for prostate cancer	Kerr et al.	BMC Cancer	2016
A Genetic Risk Score to Personalize Prostate Cancer Screening, Applied to Population Data	Huynh-Le et al.	Cancer Epidemiol Biomarkers Prev	2020
A personalised approach to prostate cancer screening based on genotyping of risk founder alleles	Cybulski et al.	Br J Cancer	2013
Association of Genomic Domains in BRCA1 and BRCA2 with Prostate Cancer Risk and Aggressiveness	Patel et al.	Cancer Res	2020
Cancer Risks Associated With Germline PALB2 Pathogenic Variants: An International Study of 524 Families	Yang et al.	J Clin Oncol	2020
Cancers associated with BRCA1 and BRCA2 mutations other than breast and ovarian	Mersch et al.	Cancer	2015
Characterization of the Cancer Spectrum in Men With Germline BRCA1 and BRCA2 Pathogenic Variants: Results From the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA)	Silvestri et al.	JAMA Oncol	2020
Clinical and histopathological characteristics of familial prostate cancer in Finland	Pakkanen et al.	BJU Int	2012
Clinical significance of PON1 L55M, Q192R and I102V polymorphisms and their association with prostate cancer risk in Polish men	Heise et al.	Pol J Pathol	2020
Clinical testing with a panel of 25 genes associated with increased cancer risk results in a significant increase in clinically significant findings across a broad range of cancer histories	Rosenthal et al.	Cancer Genet	2017
Diagnosing hereditary cancer predisposition in men with prostate cancer	Pritzlaff et al.	Genet Med	2020
Differences in cancer prevalence among CHEK2 carriers identified via multi-gene panel testing	Sutcliffe et al.	Cancer Genet	2020
Epidemiologic, clinical, and molecular characteristics of hereditary prostate cancer in Latvia	Abele et al.	Medicina (Kaunas)	2011
Ethnic disparities among men with prostate cancer undergoing germline testing	Kwon et al.	Urol Oncol	2020
Finnish Fanconi anemia mutations and hereditary predisposition to breast and prostate cancer	Mantere et al.	Clin Genet	2015
G84E germline mutation in HOXB13 gene is associated with increased prostate cancer risk in Polish men	Heise et al.	Pol J Pathol	2019
Genetic analyses supporting colorectal, gastric, and prostate cancer syndromes	Wallander et al.	Genes Chromosomes Cancer	2019
Germline genetic variants in men with prostate cancer and one or more additional cancers	Pilie et al.	Cancer	2017
Germline mutations in HOXB13 and prostate-cancer risk	Ewing et al.	N Engl J Med	2012
Germline Pathogenic Variants in 7636 Japanese Patients With Prostate Cancer and 12Â 366 Controls	Momozawa et al.	J Natl Cancer Inst	2020
Identification of Incidental Germline Mutations in Patients With Advanced Solid Tumors Who Underwent Cell-Free Circulating Tumor DNA Sequencing	Slavin et al.	J Clin Oncol	2018
Interim Results from the IMPACT Study: Evidence for Prostate-specific Antigen Screening in BRCA2 Mutation Carriers	Page et al.	Eur Urol	2019

Known susceptibility SNPs for sporadic prostate cancer show a similar association with "hereditary" prostate cancer	Cremers et al.	Prostate	2015
PALB2, CHEK2 and ATM rare variants and cancer risk: data from COGS	Southey et al.	J Med Genet	2016
Finnish Fanconi anemia mutations and hereditary predisposition to breast and prostate cancer	Seibert et al.	BMJ	2018
Population-based estimate of prostate cancer risk for carriers of the HOXB13 missense mutation G84E	MacInnis et al.	PLoS One	2013
Prevalence of Suspected Hereditary Cancer Syndromes and Germline Mutations Among a Diverse Cohort of Probands Reporting a Family History of Prostate Cancer: Toward Informing Cascade Testing for Men	Chandrasekar et al.	Eur Urol Oncol	2020
Prevalence of the HOXB13 G84E germline mutation in British men and correlation with prostate cancer risk, tumour characteristics and clinical outcomes	Kote-Jarai et al.	Ann Oncol	2015
Prostate cancer incidence in males with Lynch syndrome	Haraldsdottir et al.	Genet Med	2014
Risk of Prostate Cancer Associated With Familial and Hereditary Cancer Syndromes	Beebe-Dimmer et al.	J Clin Oncol	2020
Targeted prostate cancer screening in BRCA1 and BRCA2 mutation carriers: results from the initial screening round of the IMPACT study	Bancroft et al.	Eur Urol	2014
The role of germline mutations in the BRCA1/2 and mismatch repair genes in men ascertained for early-onset and/or familial prostate cancer	Maia et al.	Fam Cancer	2016
The spectrum of urological malignancy in Lynch syndrome	Barrow et al.	Fam Cancer	2013
Truncating and missense PPM1D mutations in early-onset and/or familial/hereditary prostate cancer patients	Cardoso et al.	Genes Chromosomes Cancer	2016

2. Not medically relevant (e.g. psychological or economic aspects) (N=13)

Title	Authors	Journal	Year
"I Am Uncertain About What My Uncertainty Even Is": Men's Uncertainty and Information Management of Their BRCA-Related Cancer Risks	Rauscher et al.	J Genet Couns	2018
Engaging Men With BRCA-Related Cancer Risks: Practical Advice for BRCA Risk Management From Male Stakeholders	Dean et al.	Am J Mens Health	2020
Genetic testing in the European Union: does economic evaluation matter?	Antonanzas et al.	Eur J Health Econ	2012
gsSKAT: Rapid gene set analysis and multiple testing correction for rare-variant association studies using weighted linear kernels	Larson et al.	Genet Epidemiol	2017
SNP interaction pattern identifier (SIPI): an intensive search for SNP-SNP interaction patterns	Lin et al.	Bioinformatics	2017
The effect of sample size on polygenic hazard models for prostate cancer	Karunamuni et al.	Eur J Hum Genet	2020
The Genetic Education for Men (GEM) Trial: Development of Web-Based Education for Untested Men in BRCA1/2-Positive Families	Peshkin et al.	J Cancer Educ	2019
Understanding of multigene test results among males undergoing germline testing for inherited prostate cancer: Implications for genetic counseling	Giri et al.	Prostate	2018
What factors may influence psychological well being at three months and one year post BRCA genetic result disclosure?	Bosch et al.	Breast	2012

What men want: Qualitative analysis of what men with prostate cancer (PCa) want to learn regarding genetic referral, counseling, and testing	Greenberg et al.	Prostate	2020
AA9int: SNP interaction pattern search using non-hierarchical additive model set	Lin et al.	Bioinformatics	2018
Development and validation of a 36-gene sequencing assay for hereditary cancer risk assessment	Vysotskaia et al.	PeerJ	2017
Development and validation of next generation sequencing based 35-gene hereditary cancer panel	Chan et al.	Hered Cancer Clin Pract	2020

3. Not specific enough (e.g. genetic pathways, carcinogenesis) (N=16)

Title	Authors	Journal	Year
Alcohol consumption and prostate cancer incidence and progression: A Mendelian randomisation study	Brunner et al.	Int J Cancer	2017
Cancer screening in Australia: future directions in melanoma, Lynch syndrome, and liver, lung and prostate cancers	Weber et al.	Public Health Res Pract	2019
Genetic cancer predisposition syndromes among older adults	Chavarri-Guerra et al.	J Geriatr Oncol	2020
Genomics of Prostate Cancer: What Nurses Need to Know	McReynolds et al.	Semin Oncol Nurs	2019
Germline testing for prostate cancer: community urology perspective	Concepcion et al.	Can J Urol	2019
Identification of genetic biomarkers in urine for early detection of prostate cancer	Kavalci et al.	Curr Probl Cancer	2020
Inequities in multi-gene hereditary cancer testing: lower diagnostic yield and higher VUS rate in individuals who identify as Hispanic, African or Asian and Pacific Islander as compared to European	Ndugga-Kabuye et al.	Fam Cancer	2019
Lung, Breast, and Prostate Cancer Patients with Unknown Ethnicity in US Department of Defense Cancer Registry Data: Comparisons to Patients with Known Ethnicity	Lin et al.	J Registry Manag	2017
New name for breast-cancer syndrome could help to save lives	Pritchard et al.	Nature	2019
Population prevalence of familial cancer and common hereditary cancer syndromes. The 2005 California Health Interview Survey	Scheuner et al.	Genet Med	2010
Prevalence and causes of elevated bone mass	Nottez et al.	Bone	2020
Publisher Correction: Shared heritability and functional enrichment across six solid cancers	Jiang et al.	Nat Commun	2019
A comparison of isolated circulating tumor cells and tissue biopsies using whole-genome sequencing in prostate cancer	Jiang et al.	Oncotarget	2015
Assessing the role of insulin-like growth factors and binding proteins in prostate cancer using Mendelian randomization: Genetic variants as instruments for circulating levels	Bonilla et al.	Int J Cancer	2016
Germline Mutations in DNA Repair Genes in Patients With Metastatic Castration-resistant Prostate Cancer	Holeckova et al.	In Vivo	2020
Microsatellite Instability Is Associated With the Presence of Lynch Syndrome Pan-Cancer	Latham et al.	J Clin Oncol	2019

4. Genetic testing in other tumor entities (N=19)

Title	Author	Journal	Year
Analysis of Founder Mutations in Rare Tumors Associated With Hereditary Breast/Ovarian Cancer Reveals a Novel Association of BRCA2 Mutations with Ampulla of Vater Carcinomas	Pinto et al.	PLoS One	2016

Application of Hereditary Renal Cell Carcinoma Risk Criteria to a Large Prospective Database	Kushnir et al.	Clin Oncol (R Coll Radiol)	2020
ATM whole gene deletion in an Italian family with hereditary pancreatic cancer: Challenges to cancer risk prediction associated with an 11q22.3 microdeletion	Arts et al.	Cancer Genet	2020
BAP1 Syndrome - Predisposition to Malignant Mesothelioma, Skin and Uveal Melanoma, Renal and Other Cancers	Foretova et al.	Klin Onkol	2019
Characteristics of individuals with breast cancer rearrangements in BRCA1 and BRCA2	Jackson et al.	Cancer	2014
Comprehensive genetic characterization of hereditary breast/ovarian cancer families from Slovakia	Konecny et al.	Breast Cancer Res Treat	2011
Germline mutation prevalence in individuals with pancreatic cancer and a history of previous malignancy	Dudley et al.	Cancer	2018
High risk men's perceptions of pre-implantation genetic diagnosis for hereditary breast and ovarian cancer	Quinn et al.	Hum Reprod	2010
Incidence of Pathogenic Variants in Those With a Family History of Pancreatic Cancer	Macklin et al.	Front Oncol	2018
Lynch Syndrome: Genomics Update and Imaging Review	Cox et al.	Radiographics	2018
Malignant Peripheral Nerve Sheath Tumor in a Patient With BAP1 Tumor Predisposition Syndrome	Kaszuba et al.	World Neurosurg	2018
Men seeking counselling in a Breast Cancer Risk Evaluation Clinic	Freitas et al.	Ecancermedicalscience	2018
Polymorphisms in MMP-1, MMP-2, MMP-7, MMP-13 and MT2A do not contribute to breast, lung and colon cancer risk in polish population	Bialkowska et al.	Hered Cancer Clin Pract	2020
Prevention and Screening in Hereditary Breast and Ovarian Cancer	Zeichner et al.	Oncology (Williston Park)	2016
Recurrent large genomic rearrangements in BRCA1 and BRCA2 in an Irish case series	McVeigh et al.	Cancer Genet	2017
Rescreening for genetic mutations using multi-gene panel testing in patients who previously underwent non-informative genetic screening	Frey et al.	Gynecol Oncol	2015
BRCA1 and BRCA2 mutations in males with familial breast and ovarian cancer syndrome. Results of a Spanish multicenter study	de Juan et al.	Fam Cancer	2015
CDKN1B V109G polymorphism a new prognostic factor in sporadic medullary thyroid carcinoma	Pasquali et al.	Eur J Endocrinol	2011
Population prevalence of individuals meeting criteria for hereditary breast and ovarian cancer testing	Greenberg et al.	Cancer Med	2019

5. Animal Studies (N=3)

Title	Authors	Journal	Year
CD24 Is Not Required for Tumor Initiation and Growth in Murine Breast and Prostate Cancer Models	Cremers et al.	PLoS One	2016
Chromosome instability and carcinogenesis: insights from murine models of human pancreatic cancer associated with BRCA2 inactivation	Cassidy et. al	Mol Oncol	2014
DNA-Encoded Library-Derived DDR1 Inhibitor Prevents Fibrosis and Renal Function Loss in a Genetic Mouse Model of Alport Syndrome	Richter et al.	ACS Chem Biol	2019

6. Case Reports (N=2)

Title	Authors	Journal	Year
Contiguous gene deletion of chromosome 2p16.3-p21 as a cause of Lynch syndrome	Salo-Mullen et al.	Fam Cancer	2018

DNA Damage Repair (DDR) Mutations and the Utility of High-Risk Genetics Clinics in Metastatic Castration-Refractory Prostate Cancer (mCRPC)	Rathi et al.	World J Oncol	2018

7. Prostate cancer but not focused on genetic testing (N=4)

Title	Authors	Journal	Year
Digitoxin Inhibits Epithelial-to-Mesenchymal-Transition in Hereditary Castration Resistant Prostate Cancer	Pollard et al.	Front Oncol	2019
Effect of family history on outcome in German patients treated with radical prostatectomy for clinically localised prostate cancer	Heck et al.	Eur J Cancer	2012
The clinical phenotype of hereditary versus sporadic prostate cancer: HPC definition revisited	Cremers et al.	Prostate	2016
Circulating Metabolic Biomarkers of Screen-Detected Prostate Cancer in the ProtecT Study	Adams et al.	Cancer Epidemiol Biomarkers Prev	2019

8. Reviews (N=16)

Title	Authors	Journal	Year
Application Areas of Traditional Molecular Genetic Methods and NGS in relation to Hereditary Urological Cancer Diagnosis	Mikhaylenko et al.	J Oncol	2020
Familial prostate cancer	Giri et al.	Semin Oncol	2016
Genetic testing for hereditary prostate cancer: Current status and limitations	Zhen et al.	Cancer	2018
Hereditary Predisposition to Prostate Cancer: From Genetics to Clinical Implications	Brandao et al.	Int J Mol Sci	2020
Hereditary prostate cancer - Primetime for genetic testing?	Heidegger et al.	Cancer Treat Rev	2019
How I Do It: Genetic counseling and genetic testing for inherited prostate cancer	Giri et al.	Can J Urol	2016
Metastatic Prostate Cancer: Effects of Genetic Testing on Care	Connors et al.	Clin J Oncol Nurs	2019
Molecular genetics and targeted therapy of WNT-related human diseases (Review)	Katoh et al.	Int J Mol Med	2017
Prostate cancer screening in BRCA and Lynch syndrome mutation carriers	Castro et al.	Am Soc Clin Oncol Educ Book	2013
Screening for familial and hereditary prostate cancer	Lynch et al.	Int J Cancer	2016
The Role of BRCA Testing in Hereditary Pancreatic and Prostate Cancer Families	Pilarski et al.	Am Soc Clin Oncol Educ Book	2019
Urological cancer related to familial syndromes	Costa et al.	Int Braz J Urol	2017
BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer	Petrucelli et al.	GeneReviews	1993
Dysregulation of the homeobox transcription factor gene HOXB13: role in prostate cancer	Decker et al.	Pharmgenomics Pers Med	2014
Genitourinary cancer predisposition syndromes	Gallagher et al.	Hematol Oncol Clin North Am	2010
Human genome meeting 2016 : Houston, TX, USA. 28 February - 2 March 2016	Srivastava et al.	Hum Genomics	2016

9. Consensus decisions (N=2)

Title	Authors	Journal	Year
Implementation of Germline Testing for Prostate Cancer: Philadelphia Prostate Cancer Consensus Conference 2019	Giri et al.	J Clin Oncol	2020

Role of Genetic Testing for Inherited Prostate Cancer Risk: Philadelphia Prostate Cancer Consensus Conference 2017	Giri et al.	J Clin Oncol	2018