

Understanding of Genetic Testing Results Among Males With Prostate Cancer: Implications for Genetic Counseling

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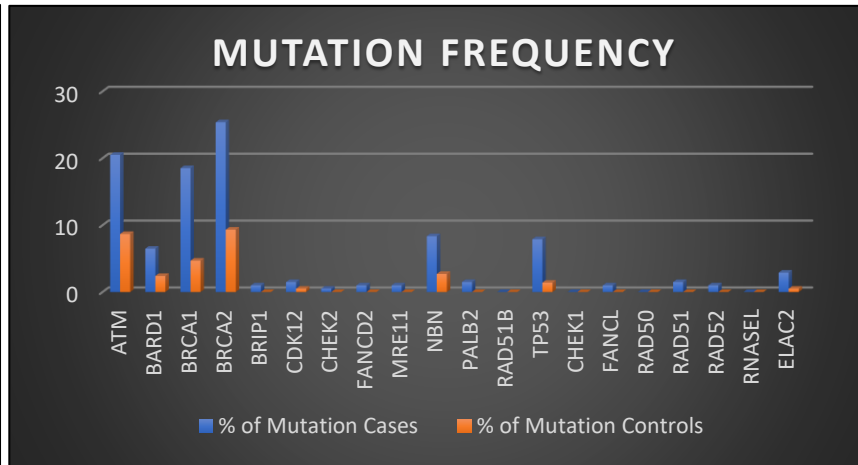


Introduction

- A significant proportion of prostate cancer diagnoses may be associated with a strong **hereditary component**.
- Approximately **8-12%** of patients with advanced prostate cancer may carry **germline mutation**.
- Genetic counseling and Germline testing** have an increasingly important role.
- Various malignancies, including **breast, ovarian, colorectal, and kidney cancers**, have been associated with hereditary syndromes.
- The emergence of **Next-Generation sequencing** has allowed for an easiest method for both somatic and germline genomic evolution.

Aim of the study is to Understand the genetic testing results among males with Prostate cancer and its implications for GC

Results



Graph 01: Mutation frequency of genes related to prostate cancer detected in the study of 40 subjects (20 Cases, 20 Controls) by NGS analysis. The most mutated genes are BRCA2, BRCA1, ATM, NBN, TP53, and BARD1.

Special Case and Genetic counseling Implications

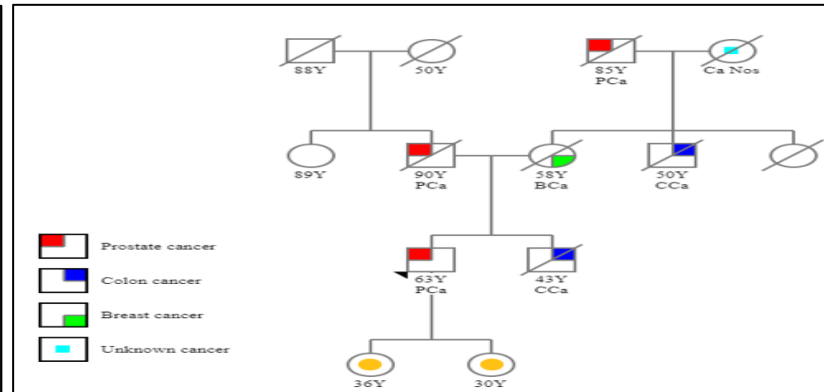
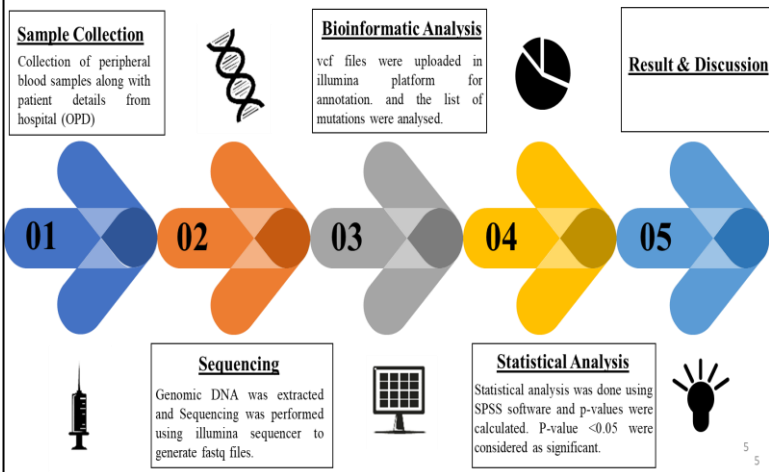


Figure 01: Proband with Pca diagnosed at the age of 63Y, Brother with Colon Cancer diagnosed at 43Y, Mother with Bca diagnosed at 58Y, and maternal uncle with Colon Cancer diagnosed at the age of 50Y.

- Family members are advised for regular screening, additional sessions for genetic counselling for the second opinion and the confirmation of clinical classification of the VUS. Especially if they have any doubt regarding the implication of the genetic test results and possible surveillance and preventive measures with their limitations.

Materials & Methods

Study group: Patients attending urology clinic with LUTS and PSA > 4ng/ml



GENE	PATHOGENIC		LIKELY PATHOGENIC		VUS	
	CASE	CONTROL	CASE	CONTROL	CASE	CONTROL
BRCA1	13	2	2	0	12	4
BRCA2	15	3	5	2	6	2
ATM	8	0	5	0	7	1
NBN	3	0	2	1	4	0
TP53	1	0	2	0	5	0
PALB2	1	0	0	1	0	0
BARD1	2	1	3	1	4	2

Table 01: No. of Common Mutations found in Cases and Controls

GENE	MUTATION TYPE	RSID	ALLELE CHANGE	CASES	CONTROLS
BRCA1	SNP	rs799916	T>A / T>C / T>G	14	01
BRCA2	Frameshift variant	rs1555284730	delGT	17	02
ATM	Frameshift variant	rs879254271	dupA	16	00
NBN	Missense Variant	rs1064795318	CA>AG / CA>TG	12	00

Table 02: List of Novel Mutations

Discussion & Conclusion

- The management of Prostate Cancer case is an evolving practice with new genetic testing guidelines from the NCCN taking into effect to deliver better patient care.
- Improvements in clinical practice and operations can lead to an increase in genetic counseling. With the changes genetic testing results can be more readily available, and ultimately enable physicians to deliver personalized care and improve clinical outcome.

References:

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