

Pathogenic variants associated with prostate cancer ethnic disparities

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Germline testing panels for prostate cancer are mainly based on studies on European ancestral populations, while **African ancestry is a significant risk factor for advanced Prostate cancer**. There is no agreement about prostate cancer germline testing of people with an African ancestry due to lack of data.

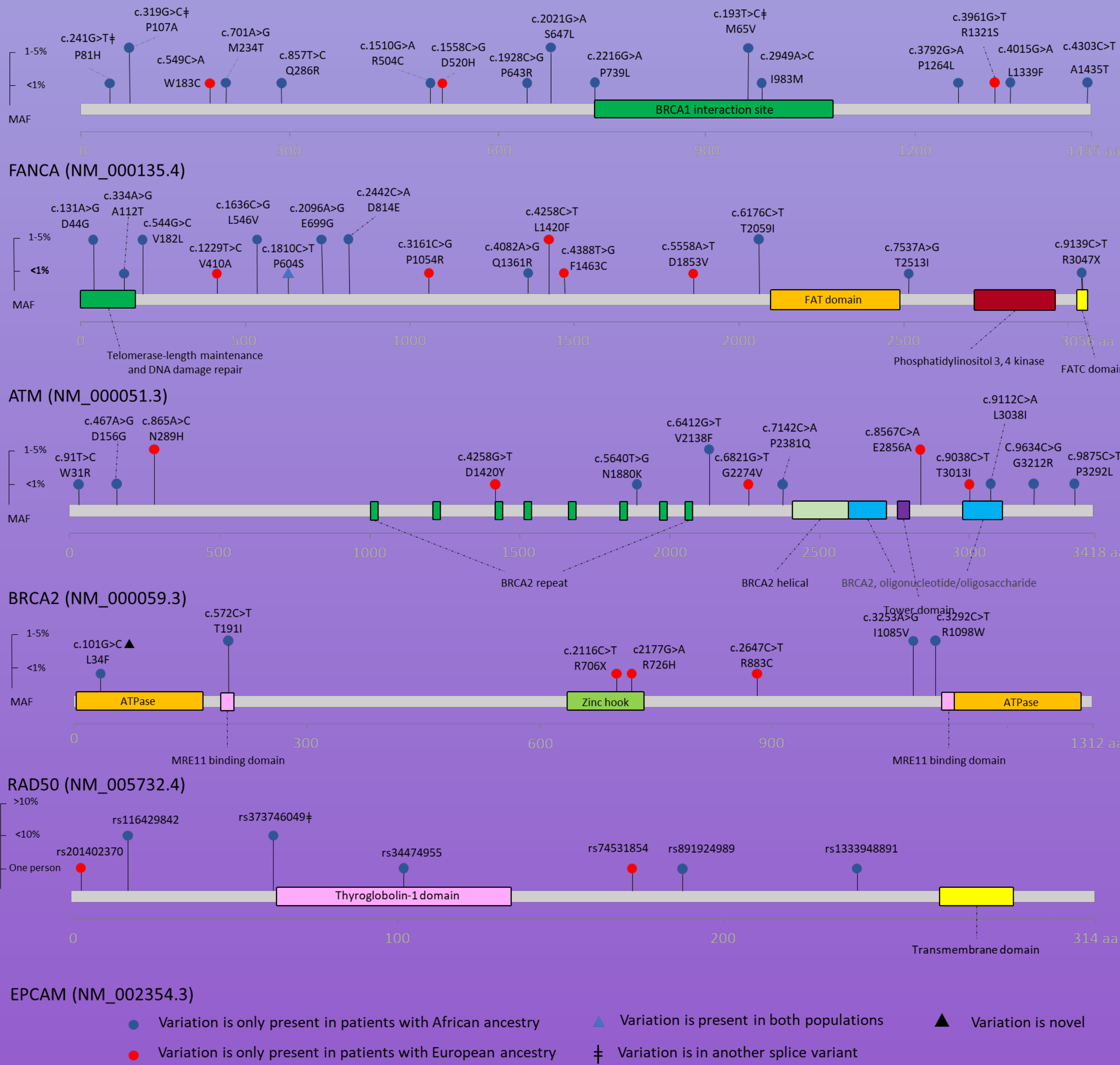


Black South African men are at 2.1-fold increased risk for advanced disease compared with African Americans. **Here we aimed to determine if current germline screening panels hold clinical benefit for men from southern Africa.**

Germline testing was performed for 20 of the most common genes in available prostate cancer genetic testing panels on 113 southern African and 53 European prostate cancer cases and the results were compared

ATM  
BRCA1  
BRCA2  
CHEK2  
EPCAM  
HOXB13  
MLH1  
MSH2  
MSH6  
NBN  
PMS2  
TP53  
BRIP1  
PALB2  
RAD51C  
RAD51D  
MUTYH  
APC  
RAD50  
FANCA

Predicted deleterious variants (PDV) in genes with higher likelihood of harbouring a variant among Africans



119 PDVs were found and only 2 were common between the two populations

**Known pathogenic variants:**  
6 for the 53 European  
vs  
4 for the 113 Africans

Known pathogenic variants were found in **ATM, TP53, BRCA2** and **CHEK2** genes in Black southern African Patients

No potentially oncogenic variants in **HOXB13, BRCA1, NBN** and **MSH2** for Africans

Several uncertain significance and 4 novel variant among Africans

Our findings showed that using the current germline testing panels for southern African patient has some merits. However, more studies need to be conducted among sub-Saharan Africans to develop germline testing panels which are more aligned to the genetic profile of this population

