My Gene Analysis:

rs1691053 A;A

rs1691053 is located on chromosome 5p15.31 and is associated with SRD5A1, which encodes the 5α-reductase isoform 1. This SNP has shown to be associated with prostate cancer (PCA), which is affected by dihydrotestosterone (DHT), the most potent male hormone. DHT functions by directly activating the androgen receptor. SRD5A along with another steroid-5α-reductase gene, HSD3B1 located on chromosome 1p12, are involved with converting testosterone to the more potent androgen DHT.

In 2010, Setlur et al. investigated the associations between SNPs in genes, HSD3B1, SRD5A1/2, and AKR1C2, that are involved in converting testosterone to DHT and known to confer PCA risk. The cohort consisted of 426 men, 205 controls and 221 cases. They were selected based on a prostate-specific antigen screening test in Tyrol, Austria. The study found that individuals with AG or GG versus AA at **rs1691053** associated with SRD5A1 have an odds ratio of 1.8 (95% CI, 1.04-3.13) with G being the risk allele. In addition, men carrying the AA genotype at rs6428830 associated with HSD3B1 have an odds ratio of 2.0 (95% CI: 1.1-4.1) compared to GG individuals. Overall, men carrying both risk alleles for SRD5A1 and HSD3B1 have an odds ratio of 3.1 (95% CI: 1.4-6.7) compared to men carrying neither with a p-value of 0.005 [PMID 20056642].

rs1691053 is one of seven SNPs identified that is associated with SRD5A1 (the others are rs566202, rs4702379, rs248803, rs3797177, rs8192249, and rs30434). In addition, eight other SNPs were identified that are associated with HSD3B1 (rs6428830 and rs10754400), SRD5A2 (rs4952197, rs7594951, and rs806645), and AKR1C2 (rs11252866, rs11252867, and rs11816204). This study by Setlur et al. is the first to determine that 5α-reductase isoform 1 is related to PCA risk and the **rs1691053** SNP might be used as a screening process in the future to decide whether to block the enzymatic activity of both isoforms of 5α-reductase for PCA chemoprevention.

Orientation	minus
Stabilized	minus

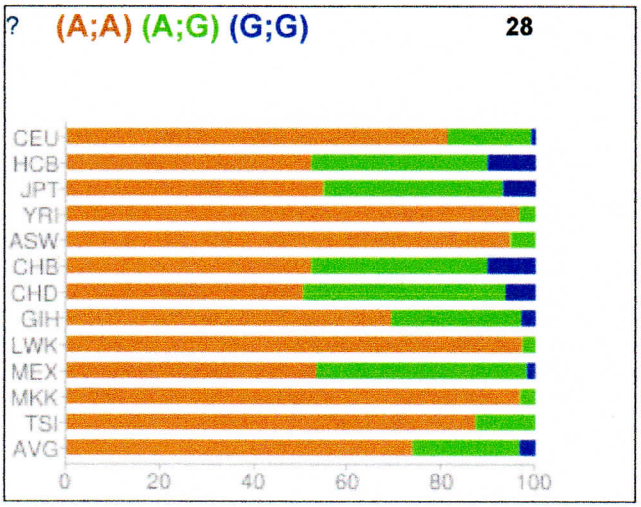
Geno	Mag	Summary
(A;A)	0	common in complete genomics
(A;G)	2	Increased risk of developing prostate cancer
(G;G)	2	Increased risk of developing prostate cancer

Reference	GRCh38 38.1/141
Chromosome	5
Position	6677052
is a	snp
is	mentioned by
dbSNP	rs1691053
dbSNP (old)	rs1691053
ClinGen	rs1691053
ebi	rs1691053
HLI	rs1691053
Exac	rs1691053
Gnomad	rs1691053
Varsome	rs1691053
Map	rs1691053
PheGenI	rs1691053
Biobank	rs1691053
1000 genomes	rs1691053
hgdp	rs1691053
ensembl	rs1691053
gopubmed	rs1691053
geneview	rs1691053
scholar	rs1691053
google	rs1691053
pharmgkb	rs1691053
gwascentral	rs1691053
openSNP	rs1691053
23andMe	rs1691053
23andMe all	rs1691053
SNPshot	rs1691053
SNPdbe	rs1691053

More recently, a research group in Spain genotyped 494 consecutive Spanish patients diagnosed with nonmetastatic localized PCA for 32 SNPs in SRD5A1, SRD5A2, and CYP17A1. They were able to analyze genotypic and allelic frequencies and haplotypes. They found that there were two SNPs in SRD5A1, rs3822430 and **rs1691053**, that are associated with prostate-specific antigen levels.

Patients who are G carriers for both SNPs compared to AA-AA carriers are at a higher risk of having high prostate-specific antigen levels of greater than 20 ng/mL with an odds ratio of 2.812 (95% CI: 1.397-5.657) and a p-value of 0.004. Through a haplotype analysis, the researchers found that patients with PCA that is nonhomozygous for the haplotype GCTTGTAGTA are at a higher risk of having a larger clinical tumor size with an odds ratio of 3.823 (95% CI: 1.280-11.416) and a p-value of 0.016 [PMID 25960412].

MSV3d	rs1691053
GWAS Ctlg	rs1691053
GMAF	0.1391
Max Magnitude	2



Categories: Is a snp | In dbSNP | SNPs on chromosome 5 | Has genotype | Has population | On chip 23andMe v3 | On chip 23andMe v4 | On chip Affy GenomeWide 6 | On chip Ancestry v2

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