

# BONE MINERAL DENSITY AND ASSOCIATED GENETIC VARIANTS IN HIGH-LEVEL CAUCASIAN MARATHON RUNNERS

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## INTRODUCTION:

- Endurance runners tend to have high total and/or loading site-specific bone mineral density (BMD) (Scofield & Hecht, 2012).
- BMD is approximately 50-85% heritable (Ralston & Uitterlinden, 2010).
- Little is known about which specific genes are involved, whether particular genotypes are sensitive to mechanical loading and the impact of such an interaction on BMD.
- This study investigated if high-level endurance runners possess enhanced BMD associated with an “advantageous” genetic predisposition, via a potential gene-training interaction.



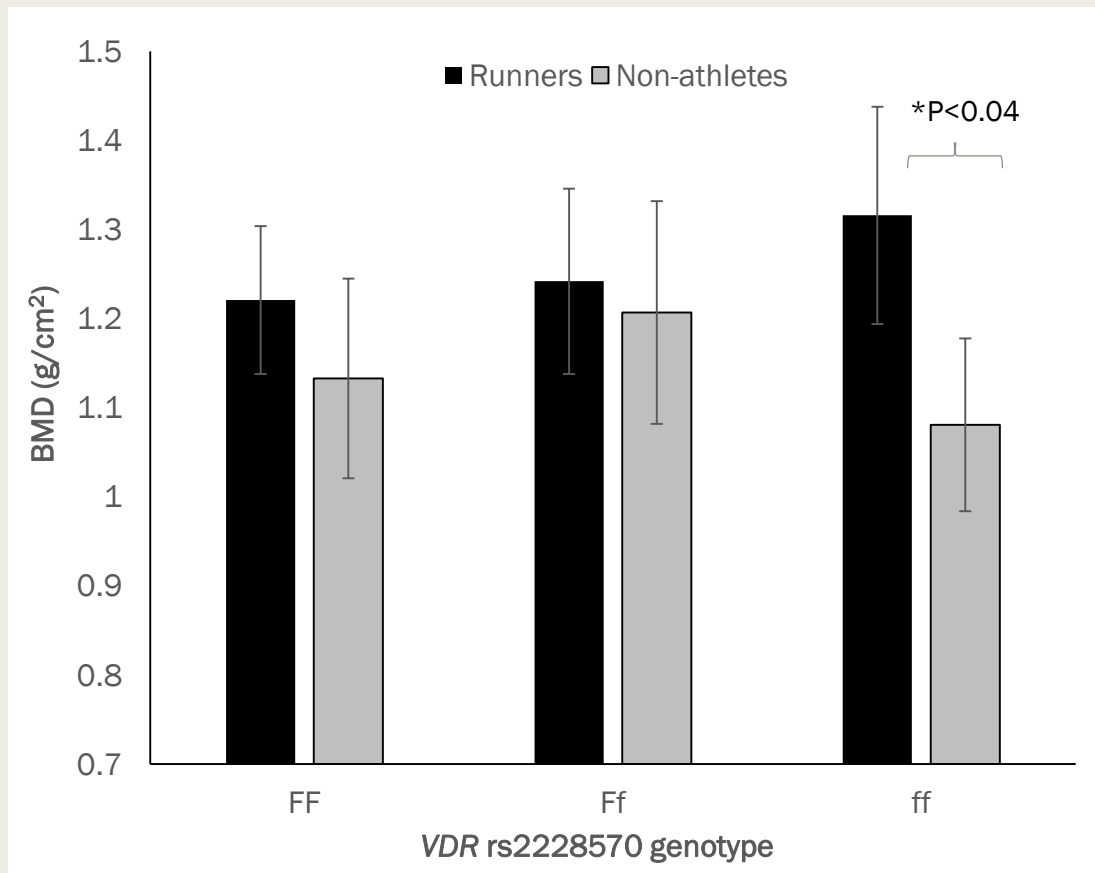


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## RESULTS:



- In males, runners had ~7% higher TBMD (1.33 vs 1.28 g/cm<sup>2</sup>;  $P < 0.05$ ) and LBMD (1.53 vs 1.43 g/cm<sup>2</sup>;  $P < 0.01$ ) than non-athletes.
- In females, runners had ~10% higher TBMD (1.24 vs 1.16 g/cm<sup>2</sup>;  $P < 0.01$ ) and LBMD (1.32 vs 1.19 g/cm<sup>2</sup>;  $P < 0.01$ ) than non-athletes.
- For *VDR* rs2228570, ff genotype was associated with higher TBMD in female runners but lower TBMD in female non-athletes suggesting an interaction between genotype and physical training load ( $*P < 0.04$ ).
- No other interactions or variants, individually or collectively as part of a TGS, were associated with BMD ( $P > 0.10$ ).

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## CONCLUSION:

- High-level male and female runners possess both higher TBMD and LBMD in comparison with non-athletes.
- Consistent with most prior literature, higher BMD was observed in *VDR* rs2228570 FF and Ff genotypes in non-athletes, which may be due to increased biological activity associated with the F variant (Arai et al., 1997; Yasovanthi et al., 2011).
- However, our preliminary data suggest the ff genotype may be associated with enhanced TBMD in female runners via interaction with the high volume of (bone stimulating) training conducted, i.e. a gene-environment interaction.