GENE VARIATIONS AND LEFT VENTRICULAR HYPERTROPHY IN ATHLETES
Ahmetov Ildus¹, Linde Elena, Hakimullina Albina, Shikhova Julia, Astratenkova Irina
(St Petersburg Research Institute of Physical Culture ¹, Russia)

Left ventricular hypertrophy (LVH) in endurance-oriented athletes is generally understood to be a limiting factor for improving aerobic performance. Studies in related and unrelated individuals clearly demonstrate that a high proportion of interindividual variability in left ventricular mass (LVM) and risk of LVH is attributable to genetic factors. Recent advances in defining the genetic variations responsible for LVH raise the possibility of DNA diagnosis in athletes. Left ventricular growth is regulated by several independent signaling pathways. These include pathways involved with the calcineurin/NFAT (calcium signaling), PGC1/TFAM (mitochondrial biogenesis, fatty acid oxidation), HIF/VEGF (vascular growth) etc. The purpose of the study was to investigate gene polymorphisms for association with LVH in athletes. Seventy one Russian athletes (all-round speed skaters and rowers) of national competitive standard (sub-elite and elite) were studied. HIF1A (hypoxia-inducible factor 1, alpha) Pro582Ser, NFATC4 (nuclear factor of activated T-cells, calcineurin-dependent 4) Ala160Gly, PGC1B (PPARgamma coactivator-1-beta) Ala203Pro, TFAM (transcription factor A, mitochondrial) Thr12Ser, VEGF (vascular endothelial growth factor) C-2578A and G-634C gene polymorphisms were determined by PCR-RLFP. Echocardiography was performed for the measurement of left ventricular mass and function. We found that LVM and LVM index (LVMI) in male sub-elite speed skaters was significantly greater in VEGF GG genotype carriers than in heterozygotes (GC) (LVM: 333 (21) g vs. 254 (21) g, P=0.002; LVMI: 169 (10) g/m² vs. 130 (18) g/m²; P=0.015). VEGF A allele carriers in elite speed skaters group had greater values of LV posterior wall thickness (LVPWd) (AA+AC – 1.53 (0.1) cm, CC – 1.2 (0.1) cm; P=0.019). In male athletes NFATC4 Ala/Ala genotype was associated with higher risk of LVH (for LVM: Ala/Ala – 398 (77) g vs. 328 (70) g; P=0.01). Furthermore, PGC1B Pro allele was found protective against LVH in both male and female speed skaters (males’ LVPWd: Ala/Ala – 1.38 (0.1) cm, Ala/Pro – 1.1 (0) cm, P=0.017; females’ LVM: Ala/Ala – 165 (14) g, Ala/Pro – 224 (37) g, P=0.06). In male rowers LVM was significantly greater in TFAM SS heterozygotes than in T allele carriers (SS – 409 (63) g, ST+TT – 324 (87) g; P=0.029). In conclusion, NFATC4, PGC1B, TFAM and VEGF gene polymorphisms are associated with risk of left ventricular hypertrophy in athletes.

Keywords: Genetics, Genotype, Athlete’s Heart

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