

Critical appraisal of sports genetics

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SUMMARY OF MAIN FINDINGS

In **chapter 2**, I addressed a critical issue in biomolecular/sports genetic research: the limitations of current sports typologies. The groundwork for this work was laid during the systematic review (chapter 3), particularly during the data extraction phase. We found that many articles identified through our systematic search strategy failed to adequately describe their athletic cohorts and/or the sport disciplines under investigation. These articles often omitted essential details such as distances, times, and performance levels, yet still applied broad labels such as endurance or strength. Sport scientists, especially those in the field of genetics, frequently classify athletic cohorts into arbitrary categories and create a practical framework for studying diverse athletic populations across seemingly similar groups. It is crucial to acknowledge the limitations and complexities of these classifications, as they tend to oversimplify the multidimensional characteristics of each sport. As a result, the validity of studies that rely on such approaches is compromised, and the comparability across different studies becomes challenging or impossible. Only with a standardized classification system large-scale meta-analysis with high-quality data become feasible, which is essential for advancing sports genetic research.

In **chapter 3**, I presented a systematic review, including meta-analyses, aimed at identifying the genetic variants of (inter)national competing long-distance runners and road cyclists compared with controls. This systematic review comprised 43 studies, including a total of 3,938 athletes and 10,752 controls in the pooled analysis. Of the 42 identified genetic variants, 13 were investigated in at least two independent studies and thus could be included in the pooled analysis. Significant associations were found for five polymorphisms. The pooled odds ratio [95%CI] favouring athletes compared with controls was 1.42 [1.12-1.81] for *ACE II* (I/D), 1.66 [1.26-2.19] for *ACTN3* TT (rs1815739), 1.75 [1.34-2.29] for *PPARGC1A* GG (rs8192678), 2.23 [1.42-3.51] for *AMPD1* CC (rs17602729), and 2.85 [1.27-6.39] for *HFE* GG + CG (rs1799945). The risk of bias was low in 25 (58%) articles and unclear in 18 (42%). Heterogeneity of the results was low (0%-20%) except for *HFE* (71%), *GNB3* (80%), and *NOS3* (76%). The findings indicate that (inter)national competing runners and cyclists have a higher probability of carrying specific genetic variants compared with controls. Ultimately, this