Olympic Genes on the Podium?

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Despite some advances, it remains largely unknown how the millions of variations in the human genome influence athletic performance (especially in endurance events), and no single genetic test can really predict sports talent. However, there is experimental evidence from animal research that selecting for even a simple characteristic such as running ability can produce comparatively large and rapid changes in performance. That such selection has not been specifically documented in humans is more evidence of the limits of physiology-archeology than of the unlikelihood of selection for physical abilities. Here, the authors argue that top Olympians are likely genetically gifted individuals who in addition have numerous contributors to the “complex trait” of being an athletic champion that may not necessarily depend on defined genetic variations.

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The Rio 2016 Olympic Games have recently concluded. The home of many champions was largely predictable. Sprinting champions are of West African descent, whereas the best endurance runners are of East African descent. Does this track and field paradigm really reflect the genetic endowment that some ethnicities or individuals are essentially predetermined, or at least are very strongly biased, to excel in certain sport specialties?

There are data supporting the concept that “speed” and “endurance” genes differ.1 During human evolution, the “trade-off” between these genes would have predisposed individuals to be more “sprinters” or more “stayers.” This phenomenon is illustrated by the R577X variation in ACTN3, a strong “speed gene” candidate encoding α-actinin-3, a protein required for explosive muscle contraction. Approximately 1 billion people worldwide (~11% of the population) are α-actinin-3-deficient because of a genetic defect in both gene copies. Yet, this “null” (XX) genotype, which is much more infrequent among Jamaicans and African Americans,2 considerably decreases the likelihood of Olympic success in sprint events.3 The R577X variation, which appeared 40,000 to 60,000 years ago in Eurasia and would have made anatomically modern humans slower and more enduring, offered a survival benefit for persistence hunters at the expense of losing muscle power.4 By contrast, it is predatory animals’ capacity for explosive muscle actions that is essential for survival.

However, despite some advances,5 it remains largely unknown how the millions of variations in the human genome influence athletic performance, and no single genetic test can reliably predict sports talent.6 A recent genome-wide exploration in 8 cohorts of world-class endurance athletes from different ethnicities and continents failed to identify a panel of genomic variants common to sports success.7 However, we have well-controlled experimental evidence in rats that selecting for even a simple characteristic such as running ability can produce comparatively large and rapid changes in anatomy, physiology, and running ability.8,9 That such selection has not been specifically documented in humans is more evidence of the limits of physiology-archeology than of the unlikelihood of selection for physical abilities.

Williams and Folland10 proposed that elite athletes are selected from individuals with a high likelihood of matching some ideal combination of genotypes in several key polymorphisms. They suggested that 23 genotypes (based on 2008 data) are required for top-level performance, assuming that an elite athlete must have all 23 genetic variants, and then the likelihood of finding a champion is a 0.0005% chance for any individual.10 However, the larger the population, the more likely it is to find an individual with all the necessary genotypes to allow phenotypic expression of the champion athlete. The historical improvement in world records over time as world population increases and as demographics change dictates that the number of people living in countries with at least potential access to high-level completion increases (eg, the emergence of East African runners in the late 1960s was more a sociocultural event, as the Kenyan and Ethiopian runners had always been there). The stagnation of world-best performances after the death of large numbers of athlete-age (more often male) individuals in war11 can also be explained by a reduction in the number of potential individuals in the selection pool. Similarly, the relative flatness of current performance quality across the athletic spectrum suggests that population-level selection for athleticism may be approaching its limits. It also depends on the sport; that is, the likelihood might be comparatively low for “overall” running (~10% of athletic individuals) and even for middle-distance running (~25% of runners), as athletic individuals are more likely drawn to team sports. Even here, the number of young adults who even attempt to participate in sport in a systematic way is limited to about 20%. Furthermore, the length of the competitive window (approximately 18–30 y in most societies, or about 16% of the life span) limits the size of the pool of potential genetically gifted competitors.

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Thus, an “equation” of the genetics of top sport can be written as

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\text{Population of the planet in 2016} = 7.3 \times 10^9
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Likelihood of inheriting a required number of “athletic genotypes” based on the Williams and Folland\textsuperscript{10} estimate = 0.000005

Percentage of people who make a systematic attempt at organized sport = 0.20

Percentage of the planet who are male = 0.48

(\text{lower in postwar periods})

Percentage of people who have access to any high-level sport = 0.9

Percentage of the planet who choose running as their sport choice = 0.1

Percentage of runners who choose middle- to long-distance running = 0.25

Percentage of the lifetime (75 y) in which top sport is likely (12 y) = 0.16

Solving this equation, we get ~13 individuals with the required genotype for top-level competition in middle- to long-distance running.

When one considers that an Olympic final will have at most 12 individuals, and if we consider the 1500-m, 3000-m steeplechase, 5000-m, and 10,000-m as representative of middle- to long-distance running, then at most there are (ignoring overlaps) 52 truly elite male middle- to long-distance runners on the planet at any given time, a number that is within 1 order of magnitude of the potential number of people on the planet (~13) capable of top-class performance. When one considers the circumstances, injuries, political and economic barriers, and selection into other sports that are likely to prevent all of the truly top “genetic candidates” from participating, the match within 1 order of magnitude of the top sport genomic equation and Olympic finalists supports the concept that the top Olympians are likely genetically gifted individuals. Moreover, they possess numerous contributors to the “complex trait” of being an athletic champion that may not depend on defined genetic variations. It seems reasonable to suggest that the genetic requirements in other sports might be different, although the presence of a large number of required genes as suggested by Williams and Folland\textsuperscript{10} is still plausible. Supporting factors required in addition to genetic predisposition include social support, opportunity, economic possibility, hard work, and motivation, all of which may be independent of gene-driven physiological factors. In the case of female athletes, the percentage of people who have access to high-level sport is markedly lower because of the history and lingering presence of factors acting to keep women out of sport.

In the end, we believe that it is likely that while watching the Olympics we are observing some of the most unique individuals on the planet. However, we agree with Wilber and Pitsiladis\textsuperscript{13} that the best evidence available today supports the concept that Olympic success is based on “favorable somatotypical characteristics lending to exceptional biomechanical and metabolic economy/efficiency; chronic exposure to altitude [eg, appropriate environment] in combination with moderate-volume, high-intensity training . . . . and a strong psychological motivation to succeed”\textsuperscript{(p92)} as on any currently definable genetic “soup.”

References


