



Genomic predictors of physical activity and athletic performance

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Contents

1. Introduction	312
2. Genomic predictors of physical activity	314
2.1 Candidate gene studies	314
2.2 Genome-wide association studies	319
2.3 Challenges and future directions	327
3. Mendelian randomisation studies on physical activity	339
4. Genes and athletic performance	341
4.1 Genetic profile of endurance athletes	345
4.2 Genetic profile of power athletes	356
4.3 Genetic profile of strength athletes	357
5. Conclusions	379
References	379

Abstract

Physical activity and athletic performance are complex phenotypes influenced by environmental and genetic factors. Recent advances in lifestyle and behavioral genomics led to the discovery of dozens of DNA polymorphisms (variants) associated with physical activity and allowed to use them as genetic instruments in Mendelian randomization studies for identifying the causal links between physical activity and health outcomes. On the other hand, exercise and sports genomics studies are focused on the search for genetic variants associated with athlete status, sports injuries and individual responses to training and supplement use. In this review, the

findings of studies investigating genetic markers and their associations with physical activity and athlete status are reported. As of the end of September 2023, a total of 149 variants have been associated with various physical activity traits (of which 42 variants are genome-wide significant) and 253 variants have been linked to athlete status (115 endurance-related, 96 power-related, and 42 strength-related).



1. Introduction

It is well established that physical activity (PA) is linked with a range of health benefits (Kraus et al., 2019; Zhang et al., 2017), and a sedentary lifestyle and/or a lack of engagement in physical activity has the opposite effect (Lee, Shiroma, & Lobelo, 2012). Interestingly, elite athletes, who exert greater amounts of physical effort than non-athletes, live ~5 years longer than the general population, an advantage mainly conferred by lower risks of cardiovascular disease and cancer (Antero, Tanaka, De Laroche Lambert, Pohar-Perme, & Toussaint, 2021). Despite the available evidence, a significant proportion of the global population fails to perform the amount of physical activity recommended to achieve positive health outcomes (Kohl et al., 2012). Variability between individuals in PA engagement is likely from multiple components, including socio-economic and cultural factors and evidence from twin and family studies that identify considerable heritability in PA phenotypes (Lightfoot, DeGeus, & Booth, 2018).

A recent review (De Geus, 2023) performed meta-analyses on twin and family studies, calculating mean heritability estimates ranging from 21% to 48% and 19% to 54%, respectively. For family studies, estimates were influenced by PA phenotype (moderate to vigorous physical activity, total physical activity, leisure time physical activity, or voluntary exercise behaviour) and measurement method (self-report versus device-based). For twin studies, the range in heritability estimates was again influenced by PA phenotype and measurement method, as well as the zygosity (monozygotic versus dizygotic) and age (<12 years, ≥12, and <18 years, or ≥18 years) of the twin pairs. Overall, de Geus estimates PA heritability to be around 43% across the lifespan, supporting previous reviews in highlighting a notable contribution of genetics to PA (De Geus, Bartels, Kaprio, Lightfoot, & Thomis, 2014; De Vilhena E Santos, Katzmarzyk, Seabra, & Maia, 2012; Lightfoot et al., 2018).

Physical activity in daily life can be categorised into occupational, sports, conditioning, household, or other activities. Exercise is a subset

of planned, structured, and repeated physical activity with a final or intermediate objective aimed at improving or maintaining physical fitness. Sports also involve physical activity and exercise but differ in that they also have rules or goals that require participants to train toward and excel in specific athletic skills. Sports at the elite level are always competitive. Athletic success is influenced by many genetically determined factors, including transcriptomic, epigenetic, biochemical, histological, anthropometric, physiological, and psychological traits, as well as general health status (Ahmetov et al., 2020; Bezuglov et al., 2023a; Blume & Wolfarth, 2019; Dines et al., 2023; Hall et al., 2021c; Rees et al., 2016; Semenova, Khabibova, Borisov, Generozov, & Ahmetov, 2019b; Stepto et al., 2009; Turner et al., 2020; Zhelankin, Iulmetova, Ahmetov, Generozov, & Sharova, 2023). On average, 66% of the variance in athlete status can be explained by genetic factors (De Moor et al., 2007). The remaining variance is due to environmental factors, such as deliberate practice, nutrition, ergogenic aids, birthplace, the availability of medical and social support, and even luck (e.g., birthdate) (Beck, Thomson, Swift, & Von Hurst, 2015; Bezuglov et al., 2023b; Macnamara, Hambrick, & Oswald, 2014; Rees et al., 2016).

In general, genetic research in the domains of physical activity, sports, nutrition, and health are interrelated and can be classified as lifestyle and behavioural genomics (Fig. 1). Additionally, research in the field of sport and exercise genomics can be divided into four main areas: genomics of athlete status and athletic performance (e.g., genetic aspects of talent identification), genomics of exercise training (e.g., individual responses to aerobic and resistance training), exercise- and sports nutrigenomics (e.g., individual responses to caffeine or protein intake), and genomics of exercise-related medical conditions (e.g., injuries, concussion, sudden cardiac death etc.) (Ahmetov, Hall, Semenova, Pranckevičienė, & Ginevičienė, 2022) (Fig. 1).

It should be noted that earlier researchers have already made attempts to systematise knowledge in the field of genetic markers that relate to exercise- and sport-related traits (Aasdahl et al., 2021; Lin, Eaton, Manson, & Liu, 2017; Moore-Harrison & Lightfoot, 2010; Semenova, Hall, & Ahmetov, 2023a). The purpose of this review is to present the latest information on genomic predictors of physical activity and athlete status.

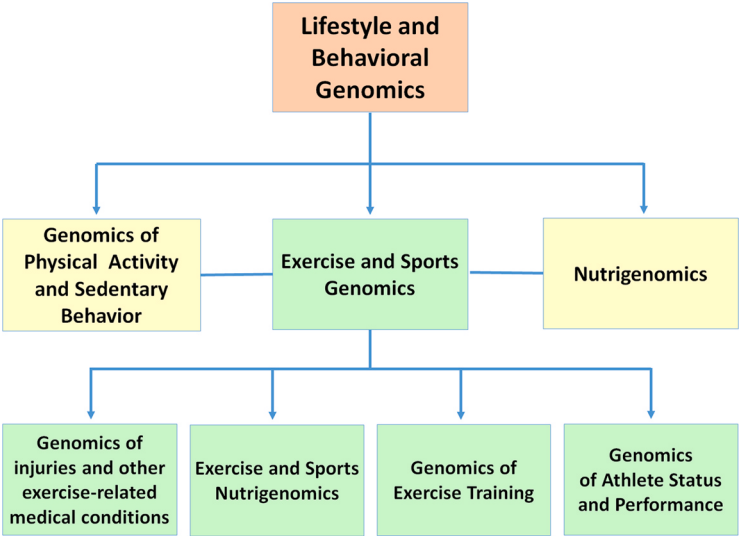


Fig. 1 Interactive branches of the lifestyle and behavioural genomics domain.



2. Genomic predictors of physical activity

There are two main types of genetic association studies: candidate gene studies and genome-wide association studies (GWAS). Both types of studies were used to discover genetic markers associated with physical activity. As of the end of September 2023, a total of 149 variants have been associated with various physical activity traits (of which 42 variants are genome-wide significant).

2.1 Candidate gene studies

Historically, most studies seeking to determine which genetic variants contribute to the heritability of PA behaviours utilised the candidate gene approach. This approach is hypothesis-driven, where specific variants within genes or genomic regions suspected to influence PA-based are investigated because of known biology. [Table 1](#) summarises candidate gene polymorphisms associated with various PA phenotypes. A common challenge of any candidate gene study is independently replicating findings in another candidate gene or Genome-Wide Association Study (GWAS).

Nevertheless, some genes have been associated with PA phenotypes in multiple studies. For example, polymorphisms in the Angiotensin-Converting

Table 1 Candidate gene associations with physical activity phenotypes.

Gene	SNP	Author (year)	PA phenotype	Association reported
<i>ACE*</i>	rs4340 (I/D)	Bruneau et al. (2017) Mäestu et al. (2013) Wong, Zhao, and Koh (2012)	Walking distance (kilometres/week) Sedentary behaviour (mins/day) Light PA (mins/day) Total PA (mins/day) Low PA PA level (kcal/week)	DD > II > ID I allele higher vs D allele D allele higher vs I allele D allele higher vs I allele D allele lower PA vs II (OR = 6.88) II > ID > DD
<i>ACTN3</i>	rs1815739	Goleva-Fjellet et al. (2020)	Low/medium vs high PA	X allele 10% less frequent in high PA group
<i>ADRB1</i>	rs1801253	Many et al. (2017)	Moderate physical activity (kcal/week)	CC higher vs CG/GG
<i>ANKRD6</i>	rs61739327	Van Devereire et al. (2012)	Moderate intensity PA (hours/week)	LL highest vs PP and PL
<i>BDNF</i>	rs1401635	Reddon et al. (2016)	PA level (sedentary, moderate, very)	C allele more active (OR = 1.08)
<i>CASR**</i>	rs1081725	Lorentzon et al. (2001)	Weight-bearing PA (hours/week)	AA higher vs SS/AS
<i>CNR1*</i>	rs6454672	Flack et al. (2019)	Moderate-to-vigorous PA (mins/day)	TT higher vs CC/CT

(continued)

Table 1 Candidate gene associations with physical activity phenotypes. (cont'd)

Gene	SNP	Author (year)	PA phenotype	Association reported
<i>FTO</i> *	rs9939603	Camps et al. (2019) Klimentidis et al. (2016)	Total activity (counts/day) Sitting (hours/day)	AT > AA > TT A allele higher time spent sitting
<i>GCKR</i>	rs780094	Espinosa-Salinas et al. (2019)	No PA vs at least one bout of any PA weekly	T allele greater exercise frequency vs CC (OR = 1.86)
<i>IL15RA</i> **	rs2228059	Bruneau et al. (2018)	Light intensity PA (hours/week)	AA more active vs CC
<i>IL-6</i>	rs1800795	Moleres et al. (2009)	Leisure time PA (none, one, >1 weekly)	GC/CC higher vs GG
<i>LEP19</i>	rs2167279	Walsh et al. (2012)	Light intensity PA (hours/week) Leisure time sports activity (kcal/week) Vigorous PA (kcal/week)	GG higher vs GA/AA GG higher vs GA/AA GG higher vs GA/AA

<i>LEPR</i> *	rs1137101	Murakami et al. (2014) Richert et al. (2007)	Light PA (mins/day) Inactive time (mins/day) Energy expended on PA (kcal/day)	RR lower vs QQ/QR RR higher vs QQ/QR GA highest, AA lowest
<i>MC4R</i> **	rs17782313 rs7242169	Lee et al. (2015) Loos et al. (2005)	MET-hours per week Moderate-to-vigorous exercise (total) Inactivity (total)	C allele 3.5% lower vs TT CC > CT > TTTT > CT > CC
<i>NPC1</i> *	rs1805081	Reddon et al. (2016)	PA level (sedentary, moderate, very)	A allele less active (OR = 0.92)
<i>NTRK2</i>	rs1211166	Reddon et al. (2016)	PA level (sedentary, moderate, very)	A allele more active (OR = 1.07)
<i>PPARD</i> **	rs2267688 rs2076167	Gielen et al. (2014)	PA (counts/day)	AG higher vs AA CC higher vs AA
<i>PPARGC1A</i> **	rs8192678	Gielen et al. (2014)	Vigorous PA (mins/day)	AA higher vs GG
<i>TMEM18</i>	rs6548328	Lee et al. (2015)	MET hours per week	T allele carries 4.1% lower vs CC
<i>UCP2</i>	rs659366	Luglio et al. (2016)	PA (METs over 6 days)	GG > GA > AA

*Gene associated with PA phenotypes in another candidate gene study (including other polymorphisms). **Gene associated with PA phenotypes in GWAS study (including other polymorphisms)

Enzyme (*ACE*) gene have been associated with different PA and sedentary behaviour phenotypes in three candidate gene studies (Bruneau et al., 2017; Mäestu et al., 2013; Winnicki et al., 2004; Wong, Zao & Koh, 2012). Similarly, several variants near or within the Melanocortin 4 Receptor gene (*MC4R*) have been associated with PA in two candidate gene studies (Lee et al., 2015; Loos et al., 2005). However, separate GWAS investigations did not report any association of variants close to or within the *ACE* nor *MC4R* genes with PA phenotypes (De Moor et al., 2009; Lin et al., 2018). Additional genes with variants associated with measures of PA and/or sedentary behaviour in two candidate gene studies include the Fat Mass and Obesity-Associated (*FTO*), dopamine D2 receptor (*DRD2*), cannabinoid receptor 1 (*CNR1*), and leptin receptor (*LEPR*) genes, as well as the *NPC1* gene encoding the Niemann-Pick C1 protein. Like *ACE* and *MC4R*, GWAS failed to confirm these loci's association with PA phenotypes.

Whilst many genes associated with PA-related phenotypes in candidate gene studies have not been replicated by GWAS, a small number have. For example, variants of the Calcium-Sensing Receptor (*CASR*) and *PAPSS2* gene, which encodes an enzyme involved in the 3'-phosphoadenosine 5'-phosphosulfate synthesis, were linked to PA behaviours in candidate gene studies (Flack, Pankey, Ufholz, Johnson, & Roemmich, 2019; Lorentzon, Lorentzon, Lerner, & Nordström, 2001) and by independent GWAS (Lin et al., 2018). Similarly, associations of different SNPs close to the aromatase (*CYP19A1*) and gamma-aminobutyric acid receptor subunit gamma-3 (*GABRG3*) genes have been replicated by GWAS (De Moor et al., 2009; Lin et al., 2018). To date, very few genetic associations with PA phenotypes have achieved replication in multiple studies, though the availability of the UK Biobank has discovered additional replications. A list of genetic markers with PA associations discovered or confirmed in the UK Biobank is provided in Table 2. Those originally identified by candidate gene study yet to be replicated include variants in the *ANKRD6*, *CNR1*, *GCKR*, *IL15-RA*, *LEP19*, *NTRK2* and *TMEM18* genes (Bruneau et al., 2018; Espinosa-Salinas et al., 2019; Flack et al., 2019; Lee et al., 2015; Reddon et al., 2016; Walsh et al., 2012). Curiously, other genes associated with PA and/or sedentary behaviours in single candidate gene studies, including *ACTN3*, *ADBR1*, *BDNF*, *IL6*, *PPARD*, *PPARGC1A* and *UCP2* (Gielen et al., 2014; Goleva-Fjellet et al., 2020; Luglio et al., 2016; Many et al., 2017; Moleres et al., 2009; Reddon et al., 2016) have been linked to sports-related phenotypes in athlete populations, suggesting there may be some effect of these loci on exercise-related attributes.

2.2 Genome-wide association studies

In contrast to the hypothesis-driven candidate gene approach, GWAS investigations sequentially screen for millions of SNPs across the genome to test their effect on a particular trait. This involves no prior gene selection on the basis of known biology, permitting unknown genomic loci to be linked to the phenotype of interest. Associated loci can then be functionally tested to discern a plausible link to the trait, and/or functional annotation methods can be used to identify patterns within GWAS results. Discussion of such methods is beyond the scope of this review but well summarised elsewhere (Li & Ritchie, 2021). Despite the recognised global burden of inactivity, at the time of writing (September 2023), the number of published GWAS of PA behaviours is limited. GWA studies typically require hundreds of thousands of participants to identify variants linked to polygenic traits and have stringent corrections for multiple testing due to the high number of SNPs involved. Indeed, several GWAS investigating PA metrics report initial associations not strong enough to survive the universally accepted genome-wide significance level of $P < 5 \times 10^{-8}$ (De Moor et al., 2009; Kim et al., 2014; Lin et al., 2018).

Recently, large-scale, long-term biomedical research initiatives such as the UK Biobank have improved the capacity to perform GWAS in larger sample sizes, yielding promise in uncovering the genetic determinants of PA and sedentary behaviours, as well as non-communicable disease phenotypes that result from inactive lifestyle (Cai et al., 2023; Semenova, Pranckevičienė, Bondareva, Gabdrakhmanova, & Ahmetov, 2023b). Two initial studies using UK Biobank participants reported associations with a range of PA and sedentary behaviour phenotypes. One reports four and three loci associated with sedentary behaviour and overall physical activity, respectively (Doherty et al., 2018), with another reporting associations of 10 loci across a range of PA measures, including overall PA, moderate-to-vigorous PA, vigorous PA, and strenuous sport/exercise (Klimentidis et al., 2018). In the latter study, two SNPs in the Exocyst Complex Component 4 (*EXOC4*) gene were associated with vigorous and moderate-to-vigorous PA, and three SNPs in the Cell Adhesion Molecule 2 (*CADM2*) gene with strenuous sport/exercise, vigorous PA and moderate-to-vigorous PA. The authors also noted that an allele of the apolipoprotein E (*APOE*) rs429358 SNP linked to the risk of Alzheimer's disease (and reduction in lifespan) was associated with higher moderate-to-vigorous PA. In the same study, one SNP in the Dpy-19-like 1 (*DPY19L1*) gene was associated with

vigorous PA, with an intergenic SNP located between *DP19L1* and the Neuropeptide S Receptor 1 (*NPRS1*) gene linked to leisure-time PA in another GWAS of Japanese participants (Hara et al., 2018). A more recent GWAS reported further association of *CADM2* variants with moderate-to-vigorous PA (Wang et al., 2022). Similarly, previous genome-wide associations linking the KAT8 Regulatory NSL Complex Subunit 1 (*KANSL1*), Synaptotagmin 4 (*SYT4*) and A-kinase anchor protein 10 (*AKAP10*) genes (Doherty et al., 2018; Klimentidis et al., 2018) to PA were supported by the most recently published GWA studies (Qi et al., 2022; Wang et al., 2022). A list of independent genomic loci associated with PA phenotypes at the genome-wide level using UK Biobank and other data is provided in Table 2.

Considering the results of published GWAS collectively, the handful of variants associated with PA-related phenotypes in more than one GWAS that merit further research include those within or close to genes including *AKAP10* and *CADM2* (Klimentidis et al., 2018; Wang et al., 2022), *KANSL1* (Doherty et al., 2018; Qi et al., 2022) and *SYT4* (Doherty et al., 2018; Klimentidis et al., 2018). Interestingly, a recent study of UK Biobank participants (Klimentidis et al., 2022) revealed that liking PA correlated with device-recorded PA, as well as self-reported vigorous PA and strenuous voluntary exercise behaviour. Four loci (of 24 identified) were identified at the genome-wide significance level as associated with these PA phenotypes, with all four previously linked to PA engagement. Specifically, *CADM2* demonstrated previous association with self-reported vigorous PA and exercise behaviour and device-based vigorous PA (Klimentidis et al., 2018), *APOE* with self-reported moderate-to-vigorous PA (Klimentidis et al., 2018), *SKIDA1* with device-based total PA (Doherty et al., 2018) and morning PA (Qi et al., 2022), and *HIST1H1D* with self-reported vigorous exercise behaviour (Klimentidis et al., 2018). This study highlights the *CADM2* gene as a prospective contributor to PA phenotypes and is particularly interesting considering the abundance of *CADM2* in brain regions involved in reward processing and addiction (Ndiaye et al., 2020). Nevertheless, further confirmatory studies are required to support and expand upon these promising findings in this and other genes.

To increase the number of genomic predictors of physical activity, genome-wide significant ($p < 5 \times 10^{-8}$) markers of other exercise-related phenotypes discovered in large cohorts of untrained subjects (for example, UK Biobank, FinnGen, etc.) can be used. Accordingly, the first stage of such an investigation is to list candidate genetic markers previously

Table 2 Genome-wide significant polymorphisms associated with physical activity traits.

Chr	Location (GRCh38)	Nearest gene	Polymorphism	Alleles	Physical activity increasing allele	Traits	P	References
3	49876964	CAMKV	rs11712056	T/C	T	Sport club/gym attendance	5.7×10^{-10}	Day et al. (2018)
3	85365141	CADM2	rs7627971	T/C	T	Sport club/gym attendance	6.2×10^{-10}	Day et al. (2018)
1	62338060	KANK4	rs17315037	T/C	T	Sport club/gym attendance	1.4×10^{-8}	Day et al. (2018)
2	136856365	THSD7B	rs13002862	C/A	A	Sport club/gym attendance	3.2×10^{-8}	Day et al. (2018)
9	119930521	MIR147A	rs5018269	A/T	T	Sport club/gym attendance	3.6×10^{-8}	Day et al. (2018)
6	33978195	MIR1275	rs2499760	C/A	A	Sport club/gym attendance	3.7×10^{-8}	Day et al. (2018)
18	43188344	SYT4	rs59499656	A/T	T	Overall activity	1.9×10^{-9}	Doherty et al. (2018)
17	46249498	KANSL1-AS1	rs2696625	A/G	G	Overall activity	3.2×10^{-12}	Doherty et al. (2018)

(continued)

Table 2 Genome-wide significant polymorphisms associated with physical activity traits. (cont'd)

Chr	Location (GRCh38)	Nearest gene	Polymorphism	Alleles	Physical activity increasing allele	Traits	P	References
10	21531721	SKIDA1	rs564819152	A/G	A	Overall activity	4.2×10^{-9}	Doherty et al. (2018)
3	10413088	SEC13	rs3836464	C/C A	CA	Active-to-sedentary transition probability	2.2×10^{-9}	Qi et al. (2022)
5	153033285	GRIA1	rs12717867	G/A	A	Light-intensity physical activity	6.2×10^{-10}	Qi et al. (2022)
3	85537753	CADM2	rs5850689	T/TG	T	Liking for bicycling	1.0×10^{-10}	Klimentidis et al. (2022)
6	27310241	POM121L2	rs16897515	C/A	C	Liking for bicycling	3.2×10^{-8}	Klimentidis et al. (2022)
1	98061858	DPYD	rs10875127	G/A	G	Liking for exercising alone	9.5×10^{-9}	Klimentidis et al. (2022)
6	26229943	HIST1H1D	rs149896934	AAAA-T/A	A	Liking for exercising alone	6.8×10^{-9}	Klimentidis et al. (2022)

19	44908684	<i>APOE</i>	rs429358	C/T	C	Liking for exercising alone	1.9×10^{-9}	Klimentidis et al. (2022)
13	98593612	<i>STK24</i>	rs9584870	C/T	C	Liking for exercising with others	7.3×10^{-9}	Klimentidis et al. (2022)
2	56387164	<i>CCDC85A</i>	rs4672114	C/G	G	Liking for exercising with others	2.4×10^{-8}	Klimentidis et al. (2022)
1	173804790	<i>DARS2</i>	rs25571	CTT/C	CTT	Liking for exercising with others	4.4×10^{-8}	Klimentidis et al. (2022)
3	85293417	<i>CADM2</i>	rs13096057	A/C	A	Liking for exercising with others	2.6×10^{-9}	Klimentidis et al. (2022)
22	41539059	<i>POLR3H</i>	rs169407	A/G	A	Liking for exercising with others	1.4×10^{-8}	Klimentidis et al. (2022)

(continued)

Table 2 Genome-wide significant polymorphisms associated with physical activity traits. (cont'd)

Chr	Location (GRCh38)	Nearest gene	Polymorphism	Alleles	Physical activity increasing allele	Traits	P	References
11	46767796	F2	rs113176495	GT/G	GT	Liking for exercising with others	4.5×10^{-8}	Klimentidis et al. (2022)
11	46369130	MDK	rs61882686	A/C	A	Liking for going to the gym	5.9×10^{-9}	Klimentidis et al. (2022)
7	114888559	MDFIC	rs112403434	A/G	G	Liking for working up a sweat	7.5×10^{-10}	Klimentidis et al. (2022)
3	18688941	SATB1	rs4645151	T/C	C	Liking for working up a sweat	8.9×10^{-9}	Klimentidis et al. (2022)
11	115273326	CADM1	rs7934107	T/C	T	Liking for working up a sweat	2.3×10^{-8}	Klimentidis et al. (2022)
6	97277152	MMS22L	rs9374462	T/C	T	Overall PA-liking	4.5×10^{-10}	Klimentidis et al. (2022)
6	26175624	H2BC6	rs9379831	A/C	A	Overall PA-liking	3.6×10^{-9}	Klimentidis et al. (2022)

10	21603559	<i>SKIDA1</i>	rs11012749	C/A	A	Overall PA-liking	6.0×10^{-9}	Klimentidis et al. (2022)
13	110875904	<i>ANKRD10</i>	rs4468472	C/T	T	Overall PA-liking	6.1×10^{-9}	Klimentidis et al. (2022)
11	22527088	<i>GAS2</i>	rs3993050	C/A	C	Overall PA-liking	4.1×10^{-8}	Klimentidis et al. (2022)
6	31373131	<i>HLA-B</i>	rs2263321	A/G	G	Overall PA-liking	7.0×10^{-9}	Klimentidis et al. (2022)
1	190557122	<i>BRINP3</i>	rs17370393	T/C	T	Overall PA-liking; Liking for going to the gym	2.1×10^{-8}	Klimentidis et al. (2022)
3	85037170	<i>CADM2</i>	rs7616022	C/T	C	Overall PA-liking; Liking for working up a sweat	3.5×10^{-11}	Klimentidis et al. (2022)
10	21550891	<i>MLLT10</i>	rs11275375	I/D	D	MVPA	4.5×10^{-12}	Wang et al. (2022)
3	84961862	<i>CADM2</i>	rs1691471	T/C	T	MVPA	1.7×10^{-19}	Wang et al. (2022)

(continued)

Table 2 Genome-wide significant polymorphisms associated with physical activity traits. (cont'd)

Chr	Location (GRCh38)	Nearest gene	Polymorphism	Alleles	Physical activity increasing allele	Traits	P	References
11	66310658	CD248	rs1625595	C/T	C	MVPA	1.9×10^{-11}	Wang et al. (2022)
6	141478397	RN7SKP106	rs13201721	T/C	T	MVPA	1.8×10^{-10}	Wang et al. (2022)
17	19903515	AKAP10	rs385301	C/T	C	MVPA	1.6×10^{-9}	Wang et al. (2022)
2	100215807	LINC01104	rs1160545	T/C	T	MVPA	1.7×10^{-9}	Wang et al. (2022)
2	10821631	PDI46	rs142601240	AT/A	AT	MVPA	2.4×10^{-9}	Wang et al. (2022)
17	52213821	CA10	rs9903845	C/A	C	MVPA	4.5×10^{-9}	Wang et al. (2022)

PA, physical activity; MVPA, Moderate-to-vigorous intensity physical activity during leisure time.

identified in GWAS of exercise-related phenotypes. For example, this may include SNPs associated with phenotypes of sedentary behaviour, handgrip strength, walking pace, lean mass, and cardiorespiratory fitness. In the second stage, GWAS summary statistics of SNPs associated with physical activity are used to search for overlaps. In this case, the significance threshold for the second stage is set at $p < 0.05$ (to reduce the likelihood of potentially important findings being overlooked). We have applied such an approach to the list of genetic markers associated with cardiorespiratory fitness ($n = 160$) (Cai et al., 2023), sarcopenia-related traits (handgrip strength, walking pace, appendicular lean mass) ($n = 78$) (Semenova et al., 2023b) and sedentary behaviour (Wang et al., 2022). It was hypothesised that the alleles associated with high cardiorespiratory fitness, low risk of sarcopenia, and decreased sedentary behaviour would also be associated with increased physical activity. Accordingly, this approach led to the discovery of an additional 85 genetic markers associated with physical activity with nominal ($p < 0.05$) significance (Table 3). However, additional studies are needed to replicate these findings.

2.3 Challenges and future directions

Initial assessment appears to reveal little consensus regarding genomic loci that contribute to PA phenotype heritability. However, several differences exist between studies relating to study design, sample size, definition, measurement type, and significance testing, which affect comparison of results. One obvious disparity between studies is the presence or absence of an a priori hypothesis and selection of specific variants, where the choice of study design may be directly influenced by sample size. The requirement for stringent multiple testing correction in GWAS means that even studies screening tens of thousands of individuals can be statistically underpowered to detect the influence of a genomic locus on a particular trait, especially one that is polygenic. In addition, GWAS can identify genes or loci with unknown biological significance, requiring substantial investigation with computational and/or molecular methods to discern their role.

Consequently, candidate gene studies are suited to smaller sample sizes, but identifying specific genes in advance assumes that a functional variant exists within that locus and that a locus has a large effect on the trait, which is not often the case (ENCODE Project Consortium et al., 2020). The recent increase in sample size available for GWA studies by projects such as the UK Biobank is encouraging, though even larger studies are required to advance this field. Importantly, particularly large samples are likely to

Table 3 Nominally significant polymorphisms associated with physical activity traits in the UK Biobank cohort.

Chr	Location (GRCh38)	Nearest gene	Polymorphism	Alleles	Physical activity increasing allele	Traits	References
3	49732599	GMPPB	rs3749237	A/G	G	Cycle	UKBB (2022) and Cai et al. (2023)
2	174682944	WIPF1	rs564190295	I/D	I	Cycle	UKBB (2022) and Cai et al. (2023)
12	65982422	LLPH	rs7136054	C/G	G	Cycle	UKBB (2022) and Cai et al. (2023)
1	66521184	SGIP1	rs17129012	C/T	T	Duration of moderate activity	UKBB (2022) and Cai et al. (2023)
4	23790032	PPARGC1A	rs142991989	A/T	A	Duration of moderate activity	UKBB (2022) and Cai et al. (2023)
7	127341680	GRM8	rs12538577	C/T	C	Duration of other exercises	UKBB (2022) and Cai et al. (2023)
12	27802363	MANSC4	rs1050288	C/T	T	Duration of other exercises	UKBB (2022) and Cai et al. (2023)
18	36635242	KIAA1328	rs8085903	C/T	C	Duration of walks	UKBB (2022) and Cai et al. (2023)

5	88705981	<i>MEF2C</i>	rs2161228	C/T	C	Duration of walks	UKBB (2022) and Cai et al. (2023)
10	110813930	<i>PDCD4</i>	rs770537744	C/CAG	CAG	Duration walking for pleasure	UKBB (2022) and Cai et al. (2023)
10	112990477	<i>TCF7L2</i>	rs61875118	C/T	T	MVPA	UKBB (2022) and Cai et al. (2023)
7	131288736	<i>MKLN1</i>	rs35680304	C/T	T	Number of days/week of moderate physical activity	UKBB (2022) and Cai et al. (2023)
2	134871960	<i>ACMSD</i>	rs35987999	C/CT	CT	Number of days/week of vigorous physical activity	UKBB (2022) and Cai et al. (2023)
4	153466445	<i>TMEM131L</i>	rs71620317	C/G	G	Number of days/week of vigorous physical activity	UKBB (2022) and Cai et al. (2023)
16	69515846	<i>NFAT5</i>	rs11866219	A/C	C	Number of days/week of vigorous physical activity	UKBB (2022) and Cai et al. (2023)
4	82323149	<i>HNRNPD</i>	rs7673637	A/G	A	Number of days/week walked 10 + minutes	UKBB (2022) and Cai et al. (2023)

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Table 3 Nominally significant polymorphisms associated with physical activity traits in the UK Biobank cohort. (cont'd)

Chr	Location (GRCh38)	Nearest gene	Polymorphism	Alleles	Physical activity increasing allele	Traits	References
5	143247691	<i>NR3C1</i>	rs373999074	T/ TTGGA	T	Number of days/week walked 10 + minutes	UKBB (2022) and Cai et al. (2023)
19	5059782	<i>KDM4B</i>	rs781155278	A/ AAGCA TAG	A	Number of days/week walked 10 + minutes	UKBB (2022) and Cai et al. (2023)
2	58734001	<i>FANCL</i>	rs17049712	C/T	C	Other exercises	UKBB (2022) and Cai et al. (2023)
4	3074876	<i>HTT</i>	rs763773724	C/ CCAG CAG	C	Other exercises	UKBB (2022) and Cai et al. (2023)
4	23949395	<i>PPARGC1A</i>	rs12501032	C/G	C	Other exercises	UKBB (2022) and Cai et al. (2023)
4	94275959	<i>HPGDS</i>	rs13131414	A/G	G	Other exercises	UKBB (2022) and Cai et al. (2023)
6	45986774	<i>CLIC5</i>	rs765227758	A/ ATCTC	ATCTC	Other exercises	UKBB (2022) and Cai et al. (2023)

7	100899267	<i>ACHE</i>	rs12705096	A/G	A	Other exercises	UKBB (2022) and Cai et al. (2023)
18	28185529	<i>CDH2</i>	rs12455124	G/T	G	Other exercises; Strenuous sports or other exercises	UKBB (2022) and Cai et al. (2023)
10	73676347	<i>SYNPO2L</i>	rs3861036	G/T	T	Sports club or gym	UKBB (2022) and Cai et al. (2023)
14	91118477	<i>RPS6K45</i>	rs769062995	A/AC	AC	Sports club or gym	UKBB (2022) and Cai et al. (2023)
12	48871970	<i>RND1</i>	rs72394060	C/ CAAAA AAA	C	Sports club or gym; Other exercises	UKBB (2022) and Cai et al. (2023)
2	184106410	<i>ZNF804A</i>	rs11454753	T/TA	T	Sports club or gym; Strenuous sports	UKBB (2022) and Cai et al. (2023)
7	135639910	<i>STMP1</i>	rs10954500	G/T	T	Sports club or gym; Strenuous sports or other exercises	UKBB (2022) and Cai et al. (2023)
1	39488876	<i>BMP8A</i>	rs375963061	A/AAT	A	Strenuous sports	UKBB (2022) and Cai et al. (2023)

(continued)

Table 3 Nominally significant polymorphisms associated with physical activity traits in the UK Biobank cohort. (cont'd)

Chr	Location (GRCh38)	Nearest gene	Polymorphism	Alleles	Physical activity increasing allele	Traits	References
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17	81398689	<i>BAHCC1</i>	rs11657730	C/T	T	Strenuous sports or other exercises; Other exercises	UKBB (2022) and Cai et al. (2023)
11	107226059	<i>GUCY142</i>	rs4754197	A/G	A	Strenuous sports or other exercises; Vigorous physical activity	UKBB (2022) and Cai et al. (2023)
17	62915351	<i>TANC2</i>	rs62076571	A/G	A	Time spent doing light physical activity	UKBB (2022) and Cai et al. (2023)
19	44908822	<i>APOE</i>	rs7412	C/T	C	Vigorous physical activity	UKBB (2022) and Cai et al. (2023)
5	148658593	<i>HTR4</i>	rs9325110	C/T	C	Vigorous physical activity; Strenuous sports	UKBB (2022) and Cai et al. (2023)
6	51967696	<i>PKHD1</i>	rs13205180	C/T	C	Walking for pleasure	UKBB (2022) and Cai et al. (2023)

19	18280361	<i>JUND</i>	rs7249	T/C	T	Duration of other exercises	UKBB (2022) and Semenova et al. (2023b)
1	185168496	<i>SWT1</i>	rs10797999	T/C	T	MVPA	UKBB (2022) and Semenova et al. (2023b)
16	69833932	<i>WWP2</i>	rs4985445	A/G	A	MVPA	UKBB (2022) and Semenova et al. (2023b)
11	47644134	<i>MITCH2</i>	rs11039324	G/A	G	Number of days/week of moderate physical activity	UKBB (2022) and Semenova et al. (2023b)
5	135038418	<i>PITX1</i>	rs4976261	G/C	G	Number of days/week of vigorous physical activity	UKBB (2022) and Semenova et al. (2023b)
2	41461300	<i>PKDCC</i>	rs3035165	T/TTA	T	Other exercises	UKBB (2022) and Semenova et al. (2023b)
6	35427233	<i>PPARD</i>	rs3734254	T/C	T	Other exercises	UKBB (2022) and Semenova et al. (2023b)
9	96477559	<i>HABP4</i>	rs6477489	C/A	C	Other exercises	UKBB (2022) and Semenova et al. (2023b)

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Table 3 Nominally significant polymorphisms associated with physical activity traits in the UK Biobank cohort. (cont'd)

Chr	Location (GRCh38)	Nearest gene	Polymorphism	Alleles	Physical activity increasing allele	Traits	References
13	50537219	<i>DLEU1</i>	rs3116602	T/G	T	Other exercises	UKBB (2022) and Semenova et al. (2023b)
6	31624141	<i>PRRC2A</i>	rs2260051	A/T	A	Strenuous sports	UKBB (2022) and Semenova et al. (2023b)
9	89593312	<i>GADD45G</i>	rs1329733	A/G	A	Strenuous sports	UKBB (2022) and Semenova et al. (2023b)
6	97981461	<i>MMS22L</i>	rs9320823	T/C	T	Strenuous sports	UKBB (2022) and Semenova et al. (2023b)
7	99529017	<i>ZKSCAN5</i>	rs3843540	C/T	C	Strenuous sports	UKBB (2022) and Semenova et al. (2023b)
4	3240118	<i>HTT</i>	rs362307	C/T	C	Strenuous sports	UKBB (2022) and Semenova et al. (2023b)
17	48963885	<i>GIP</i>	rs4794005	A/G	A	Strenuous sports	UKBB (2022) and Semenova et al. (2023b)
15	74044292	<i>PML</i>	rs5742915	C/T	C	Strenuous sports or other exercises	UKBB (2022) and Semenova et al. (2023b)

2	105368167	<i>FHL2</i>	rs55680124	C/T	C	Strenuous sports or other exercises	UKBB (2022) and Semenova et al. (2023b)
4	139942211	<i>MAML3</i>	rs57800857	C/A	C	Strenuous sports or other exercises	UKBB (2022) and Semenova et al. (2023b)
6	32420769	<i>BTNL2</i>	rs2213581	T/C	T	Strenuous sports or other exercises	UKBB (2022) and Semenova et al. (2023b)
10	63431885	<i>JMJD1C</i>	rs7924036	T/G	T	Strenuous sports or other exercises	UKBB (2022) and Semenova et al. (2023b)
3	71617912	<i>FOXP1</i>	rs830643	A/G	A	Strenuous sports or other exercises	UKBB (2022) and Semenova et al. (2023b)
20	35437976	<i>GDF5</i>	rs143384	G/A	G	Time spent doing moderate physical activity	UKBB (2022) and Semenova et al. (2023b)
8	125486586	<i>TRIB1</i>	rs4870941	G/C	G	Vigorous physical activity	UKBB (2022) and Semenova et al. (2023b)
12	109445772	<i>MYO1H</i>	rs7969719	C/T	C	MVPA	Wang et al. (2022)
18	37599845	<i>CELF4</i>	rs12962050	A/G	A	MVPA	Wang et al. (2022)
4	3303389	<i>RGS12</i>	rs743699	A/G	A	MVPA	Wang et al. (2022)

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Table 3 Nominally significant polymorphisms associated with physical activity traits in the UK Biobank cohort. (cont'd)

Chr	Location (GRCh38)	Nearest gene	Polymorphism	Alleles	Physical activity increasing allele	Traits	References
7	99520454	ZKSCAN5	rs11419307	A/T/A	A/T	MVPA	Wang et al. (2022)
9	137357006	EXD3	rs73581580	G/A	G	MVPA	Wang et al. (2022)
7	133820338	EXOC4	rs13235840	A/T	A	MVPA	Wang et al. (2022)
1	111738368	INKA2	rs197439	A/G	A	MVPA	Wang et al. (2022)
6	35395071	PPARD	rs6457816	T/C	T	MVPA	Wang et al. (2022)
7	111540388	IMMP2L	rs2529484	G/C	G	MVPA	Wang et al. (2022)
2	214502435	IVWC2L	rs36079846	T/C	T	MVPA	Wang et al. (2022)
15	77732464	LINGO1	rs56151256	C/A	C	MVPA	Wang et al. (2022)
11	88842797	GRM5	rs1391954	G/T	G	MVPA	Wang et al. (2022)
1	77501822	AK5	rs71658797	T/A	T	MVPA	Wang et al. (2022)
20	63786890	ZBTB46	rs6010651	C/A	C	MVPA	Wang et al. (2022)
11	66222968	PACS1	rs4483592	C/T	C	MVPA	Wang et al. (2022)

9	131848095	MED27	rs566556664	I/D	I	MVPA	Wang et al. (2022)
20	44896842	YWHA8	rs139900206	T/TTA	T	MVPA	Wang et al. (2022)
16	31054597	ZNF668	rs4889530	A/T	A	MVPA	Wang et al. (2022)
4	17921190	LCORL	rs548236486	A/AT	A	MVPA	Wang et al. (2022)
5	161674609	GABRA6	rs1947066	G/A	G	MVPA	Wang et al. (2022)
7	140476796	MKRN1	rs17621391	C/T	C	MVPA	Wang et al. (2022)
12	117087430	TESC	rs2173650	T/G	T	MVPA	Wang et al. (2022)
1	153947810	CRTC2	rs8450	A/G	A	MVPA	Wang et al. (2022)
2	192881557	PCGEM1	rs12617870	G/T	G	MVPA	Wang et al. (2022)
4	102267552	SLC39A8	rs13107325	C/T	C	MVPA, strenuous sports	Wang et al. (2022)

include increasing heterogeneity of geographic ancestries, and efforts should be made to account for such differences.

Another challenge encountered when quantifying PA phenotypes is the method of measurement. Self-report accounts for the majority of PA recording due to the relative ease of collection, though this method is shown to have poor validity versus accelerometry (Gupta et al., 2017; Pedersen, Kitic, Bird, Mainsbridge, & Cooley, 2016) and greater measurement error (De Geus, 2023). The financial and practical implications of hundreds of thousands of participants wearing an accelerometer or other device should be considered and are likely to explain why samples with self-report data are considerably larger than those collected via device-based methods. It is interesting to note that heritability estimates of PA for family and twin studies are both higher when calculated using device-based measures versus self-report (De Geus, 2023), which may also be influenced by the documented disparity between self-reported and actual PA behaviours. Despite the historical challenges of collecting device-based PA data, smartphones may improve the ability to collect activity data at a low cost and have been shown to implement PA interventions effectively (Emberson et al., 2021). If ethical considerations around data sharing and privacy are addressed, smartphones may help improve the precision of PA data in future genetic investigations by increasing the available sample size for device-based activity recording.

Perhaps the most evident discrepancy between studies quantifying PA and sedentary behaviours, regardless of the independent variable or intervention, is the range of different activity types and definitions within and between studies. Variables reported include, but are not limited to, inactivity (Loos et al., 2005), sedentary time (Doherty et al., 2018), sitting hours per day (Klimentidis, Arora, Chougule, Zhou, & Raichlen, 2016), overall activity (Doherty et al., 2018), total leisure-time PA measured in METs (Lin et al., 2018), regular leisure-time PA (defined as 4-MET hours per week) (Hara et al., 2018), light-intensity physical activity hours per week (Bruneau et al., 2018), moderate-to-vigorous PA (Klimentidis et al., 2018) which can also be measured in minutes per day (Flack et al., 2019), vigorous PA (Klimentidis et al., 2018), strenuous sports/exercise (Klimentidis et al., 2018), physical activity level (sedentary vs. moderate active vs. very active) (Reddon et al., 2016), regular exercisers vs. non-exercisers (De Moor et al., 2009), total activity counts per day (Camps, Verhoef, Bouwman, Mariman, & Westerterp, 2019), and sports club/gym attendance (Day, Ong, & Perry, 2018). Evidently, different definitions of

what constitutes the PA phenotype make comparison between studies difficult, whereby it is plausible that some discrepancy and/or lack of replication between studies may be due to a genomic region influencing a specific PA phenotype that is not measured by another study. For example, comparing overall activity does not differentiate between the intensity of activity (i.e., light, moderate, or vigorous), nor can the proportions of moderate and vigorous intensity within a combined “moderate-to-vigorous PA” phenotype be discerned when studies adopt this approach. It is beyond the scope of this review to evaluate PA phenotype definitions and to suggest the most appropriate classification system for future studies. Nevertheless, those seeking to understand the genetic contribution to PA should remain cognisant that there is considerable heterogeneity in how PA is defined within the existing literature.



3. Mendelian randomisation studies on physical activity

Until recently, only randomised controlled trials (RCTs) could determine causal links between physical activity and health outcomes. The situation changed with the discovery of genomic predictors of exercise- and health-related traits and the widespread implementation of Mendelian randomisation (MR) studies. Mendelian randomisation is a term that applies to genetic variation to address causal questions about how modifiable exposures (e.g., physical activity, educational attainment, income, etc.) influence different outcomes (e.g., obesity, cancer, longevity etc.). The principles of MR are based on Mendel’s laws of inheritance and instrumental variable estimation methods, which enable the inference of causal effects in the presence of unobserved confounding ([Sanderson et al., 2022](#)). Recent advances in genome-wide association studies and the increasing availability of publicly available summary data on associations of genetic variants with risk factors and disease outcomes in large sample sizes have enabled powerful Mendelian randomisation analyses to be performed relatively quickly and simply ([Burgess et al., 2023](#)). The results of MR studies are expressed as odds ratios (OR) with corresponding 95% confidence intervals (CI).

The following terms are commonly used in MR studies:

- a) instrumental variable (IV): a variable associated with an exposure that is not associated with the outcome through any other pathway; a genetic

variant can be considered as an instrumental variable for a given exposure if it satisfies the instrumental variable assumptions: (1) it is associated with the exposure, (2) it is not associated with the outcome due to confounding pathways, and (3) it does not affect the outcome except potentially via the exposure (Burgess et al., 2023);

- b)** confounder: a trait that influences both the exposure and outcome of interest (e.g., educational attainment affects both physical activity and longevity (Klimentidis et al., 2018; Ni et al., 2023));
- c)** bidirectional: an effect that acts in both directions between a pair of traits so that changing one will change the other; some examples: (1) a high level of intelligence contributes to higher education, and a high level of education increases the level of intelligence (Anderson et al., 2020); (2) high physical activity leads to lower BMI, and high BMI leads to lower physical activity (Wang et al., 2022);
- d)** reverse causality: the phenomenon where an association in the direction of a hypothesised causal relation between a risk factor and outcome is observed but is due (at least in part) to prevalent outcome influencing the risk factor; for example, high physical activity does not prevent chronic back pain, but patients experiencing chronic back pain decrease their level of physical activity (Gao et al., 2021);
- e)** pleiotropy: the phenomena of genetic variants being associated with multiple phenotypes (e.g., a specific allele for a particular SNP may be associated with low level of physical activity, slow walking pace, low fat-free mass, low strength, unhealthy diet, neuroticism, and obesity (Semenova et al., 2023b)); pleiotropy can be horizontal (when genetic variant is associated with multiple phenotypes on different pathways) and vertical (when genetic variant is associated with multiple phenotypes on the same pathway).

Where possible, different genetic data sources for MR studies should be selected to minimise the chance of sample overlap in the exposure and outcome data sets (i.e., to avoid any potential biases due to sample overlap). Such types of studies are called two-sample MR studies. In some cases, mediation analysis can be added to explain the pathway(s) through which an exposure affects an outcome. For example, the effect of sedentary behaviour on myocardial infarction and heart failure can be partly mediated by body mass index (Yang et al., 2022). Two approaches can be used for estimating mediation analysis with MR: multivariable MR and two-step MR (Carter et al., 2021).

Generally, summary statistics from genome-wide association studies establish a causal relationship between physical activity and health-related traits. Accordingly, MR studies have shown a causal protective effect of increased physical activity against many diseases, such as obesity/increased BMI (Wang et al., 2022), type 2 diabetes mellitus (Yuan et al., 2023), sarcopenia (Liu et al., 2023b), low bone mineral density (Xu et al., 2021), low parental age at death (Wang et al., 2022), nonalcoholic liver disease (Zhang, Chen, Yin, Qian, & Liu, 2023), Crohn's disease (Saadh et al., 2023), sports injuries of the ankle/foot and lumbar spine (Bi, Yang, & Jiang, 2023), ankylosing spondylitis (Hu, Xing, Wang, Zhang, & Xu, 2022), female pelvic organ prolapse (Liu, Wu, Xiang, & Yuan, 2023a), chronic kidney disease (Park et al., 2022), urolithiasis (Fang et al., 2023), gallstones (Shi et al., 2023), major depressive disorder (Zheng et al., 2023a), addictive behaviours such as smoking (Iob et al., 2023), Alzheimer's disease (Zhang et al., 2022), bipolar disorder (Sun et al., 2020), migraine (Zheng, Shi, Liang, Lu, & Chen, 2023b), multiple sclerosis (Li et al., 2022), breast cancer (Dixon-Suen et al., 2022), colorectal cancer (Zhang et al., 2021), prostate cancer (Kazmi et al., 2020), severe COVID-19 (Baranova, Cao, Teng, & Zhang, 2023), myocardial infarction (Zhuo, Zhao, Chen, & Lu, 2021), deep vein thrombosis (Hu, Li, & Yang, 2023), and hypertension (Doherty et al., 2018).

Interestingly, MR studies also indicate that education (Kari et al., 2020), zinc levels (Lyu et al., 2023), low BMI (Wang et al., 2022), and absence of chronic back pain (Gao et al., 2021) may be positive factors leading to higher physical activity and thus promoting global health. Overall, these findings may guide therapeutic decision-making in clinical practice.



4. Genes and athletic performance

Starting in the late 1990s, research began to identify DNA polymorphisms associated with predisposition to certain types of sports and exercise-related phenotypes, with an initial focus on variants of the *ACE*, *ACTN3*, *AMPD1*, *PPARA*, *PPARD*, and *PPARGC1A* genes (Akhmetov, Astranenkova, & Rogozkin, 2007a; Akhmetov et al., 2007b; RiBray et al., 2009; Gayagay et al., 1998; Lucia et al., 2005a; Montgomery et al., 1998; Nazarov et al., 2001; Rubio et al., 2005; Yang et al., 2003; vera et al., 1997a; Ginevičienė et al., 2022). Initially, most research was conducted using the candidate gene approach (Cagnin, Chemello, & Ahmetov, 2019;

Maciejewska-Skrendo, Sawczuk, Ciężczyk, & Ahmetov, 2019; Massidda, Miyamoto, Beckley, Kikuchi, & Fuku, 2019b; Semenova, Fuku, & Ahmetov, 2019a; Valeeva, Ahmetov, & Rees, 2019; Varillas-Delgado et al., 2022a), which limited progress in the discovery of new genetic markers associated with exercise- and sport-related phenotypes (Wang et al., 2013b, 2016). In addition to the fact that this approach studies only a single genetic variant in isolation, most candidate gene studies in the field of sports genomics are limited by sample size. This is a potential source of type I error (false positive findings), underpinning why replicating positive associations in independent cohorts is essential. Conversely, the genome-wide association approach is considered the most powerful and efficient study design thus far in identifying genetic variants associated with complex human traits. Indeed, the application of this approach has facilitated the discovery of hundreds of single nucleotide polymorphisms (SNPs) directly or indirectly associated with exercise and sport, such as height (12,111 SNPs) (Yengo et al., 2022), appendicular lean mass (1059 SNPs) (Pei et al., 2020), testosterone levels (855 SNPs) (Ruth et al., 2020), handgrip strength (170 SNPs) (Jones et al., 2021; Tikkanen et al., 2018; Willems et al., 2017), sarcopenia (78 SNPs) (Semenova et al., 2023b), and brisk walking (70 SNPs) (Timmins et al., 2020).

It is important to consider that the discovery of 12,111 independent genome-wide ($p < 5 \times 10^{-8}$) significant SNPs associated with height (which in combination account for 40% of phenotypic variance) required the study of 5.4 million individuals (Yengo et al., 2022). Accordingly, such findings indicate that each DNA locus is likely to explain only a very small proportion of the reported phenotypic variance ($\sim 0.0033\%$). This also suggests that very large samples of athletes and non-athletic controls are needed to detect a substantial number of genetic markers associated with athlete status, with some suggesting those markers be used as part of talent identification strategies. To date, five genome-wide association (GWAS) (Ahmetov et al., 2015; Al-Khelaifi et al., 2020; Pickering et al., 2019; Rankinen et al., 2016; Semenova et al., 2022), one whole genome sequencing (WGS) (Boulygina et al., 2020) and one exome-wide association (EWAS) (Bulgay et al., 2023) studies involving athletic cohorts have been conducted. However, due to their limited sample sizes, these investigations and subsequent replication studies have resulted in the identification of only 13 genetic markers. Six markers were associated with endurance athlete status (*CDKN1A* rs236448 A, *GALNTL6* rs558129C, *NFIA-AS2* rs1572312C, *MYBPC3* rs1052373 G, *SIRT1* rs41299232 G,

and *TRPM2* rs1785440 G alleles) (Ahmetov et al., 2015; Al-Khelaifi et al., 2020; Bulgay et al., 2023; Malczewska-Lenczowska et al., 2022; Rankinen et al., 2016; Semenova et al., 2022), five with sprinter/strength athlete status (*AGRN* rs4074992C, *CDKN1A* rs236448C, *CPNE5* rs3213537 G, *NUP210* rs2280084 A and *GALNTL6* rs558129 T alleles) (Bulgay et al., 2023; Díaz Ramírez et al., 2020; Guilherme et al., 2021b; Pickering et al., 2019) and two with reaction time in wrestlers and athlete status in combat/team sports (*APC* rs518013 A and *LRRN3* rs80054135 T alleles, respectively) (Boulygina et al., 2020).

As well as the phenotypes of athlete status or competitive performance, several recent studies have investigated a broader range of traits that may relate directly or indirectly to athletic capability. These include flexibility, coordination, cardiorespiratory fitness, testosterone levels, spatial ability, stress resilience, mental toughness, fat loss efficiency, and cardiovascular and metabolic responses to training, amongst others (Ahmetov, Donnikov, & Trofimov, 2014a; Baranova et al., 2017; Bojarczuk et al., 2022; Kleyjer et al., 2022; Leońska-Duniec, Ahmetov, & Zmijewski, 2016; Leońska-Duniec et al., 2018; Leonska-Duniec, Cięższyk, & Ahmetov, 2019; Massidda et al., 2019b; Mazur et al., 2020; McAuley et al., 2022; McAuley et al., 2023; Mountford, Hill, Barnett, & Newbury, 2021; Peplonska et al., 2019; Williams et al., 2021; Yusof, Aziz, & Muhamed, 2019; Zhao et al., 2020). For example, mixed martial arts (MMA) fighters are more likely than non-athletes to carry the warrior (*COMT* rs4680 GG; Val/Val; high activity of catechol-O-methyltransferase; low dopamine) genotype (Tartar et al., 2020), whilst chess players demonstrate an increased frequency of an allele linked to improved memory and spatial ability (*KIBRA* rs17070145 T) (Ahmetov et al., 2023). Such discoveries demonstrate the broadening nature of sports genomics in recent times, with focus expanding from the traditional domain of investigating what makes elite performers different from the general population into other domains, such as sports nutrigenetics (Guest, Corey, Vescovi, & El-Sohehy, 2018; Guest, Corey, Tyrrell, & El-Sohehy, 2022; WoGuntoro Aji et al., 2023; Hall et al., 2022b; Loy, O'Connor, Lindheimer, & Covert, 2015; Muñoz et al., 2020; Rahimi et al., 2023; Rahimi, 2019; Sicova, Guest, Tyrrell, & El-Sohehy, 2021; Thomas et al., 2017; Wong et al., 2021; mack et al., 2012) and areas of sports medicine, such as genomic variants associated with soft-tissue injuries and sports-related concussion. (Antrobus et al., 2021; Antrobus et al., 2022; RaBrazier et al., 2023a; Brazier et al., 2023b; hiDe Almeida et al., 2022a; Ginevičienė & Urnikytė, 2022; Ginevičienė et al., 2022; Hall et al., 2022a; Jacob et al., 2022;

Kim et al., 2021; Kumagai et al., 2022; Varillas-Delgado, Gutierrez-Hellín, & Maestro, 2023; Rahim, Collins, & September, 2016).

Technological advancement has lowered the cost of conducting genomic studies, increasing accessibility to researchers who wish to investigate the genetic underpinnings of sport and exercise phenotypes. Consequently, sports genomics is a dynamic and continually developing field, making it important to appraise the contribution of recent advances to the field regularly. Therefore, the aim of the current review was to summarise recent progress in understanding the genetic determinants of athlete status and to detail novel DNA polymorphisms that may underpin differences between individuals in their athletic potential.

At the time of writing (end of September 2023), the total number of DNA polymorphisms associated with athletic performance since the first in 1998 is 253. In recognition of the fact that many studies in the field of sports genomics report associations based on the investigation of small sample sizes, we stipulated that only markers where statistically significant associations have been reported in *at least two studies* (two case-control studies and/or one case-control plus one functional study; including those presented in one article) would be included in the present review.

According to these criteria, 128 markers are associated with athlete status (41 endurance-related, 45 power-related, and 42 strength-related) from the original 253 identified in our literature search. The most promising genetic markers include *AMPD1* rs17602729C, *CDKN1A* rs236448 A, *HFE* rs1799945 G, *MYBPC3* rs1052373 G, *NFIA-AS2* rs1572312C, *PPARA* rs4253778 G, and *PPARGC1A* rs8192678 G alleles for endurance; *ACTN3* rs1815739C, *AMPD1* rs17602729C, *CDKN1A* rs236448C, *CPNE5* rs3213537 G, *GALNTL6* rs558129 T, *IGF2* rs680 G, *IGSF3* rs699785 A, *NOS3* rs2070744 T and *TRHR* rs7832552 T alleles for power; *ACTN3* rs1815739C, *AR* ≥ 21 CAG repeats, *LRPPRC* rs10186876 A, *MMS22L* rs9320823 T, *PHACTR1* rs6905419C, and *PPARG* rs1801282 G alleles for strength. The current review lists all known markers associated with endurance, power, or strength athlete status/performance. This review does not aim to review genetic markers associated with team (game) and combat sports, markers for which are well described elsewhere (Demirci et al., 2023; Egorova et al., 2014; Massidda et al., 2019a; McAuley et al., 2021; Murtagh et al., 2023; Youn, Ko, & Kim, 2021).

4.1 Genetic profile of endurance athletes

Endurance performance is a complex phenotype influenced by a multitude of factors, many relating to cellular metabolism and cardiovascular function. Specifically, an individual's endurance capacity is determined by their muscle fibre typology (proportion of slow-twitch fibres in skeletal muscle), haemoglobin mass, maximal cardiac output, and maximal rate of oxygen consumption ($\text{VO}_{2\text{max}}$), among others (Abernethy, Thayer, & Taylor, 1990; Ahmetov, Vinogradova, & Williams, 2012; Bassett and Howley, 2000; Konopka et al., 2022; Malczewska-Lenczowska et al., 2016; Semenova et al., 2019a; Simoneau & Bouchard, 1995). Indeed, there is evidence that these intermediate phenotypes have a substantial genetic influence, with literature indicating that genetic factors account for 44–68% of the variability in endurance-related phenotypes (Miyamoto-Mikami et al., 2018). Traditionally, genetic variants associated with endurance athlete status are determined by comparing allelic frequencies between endurance athletes (e.g., marathon runners, 800–1500 m swimmers, triathletes, rowers etc.) and non-athletic controls. To support the observed findings from endurance-related case-control studies, researchers subsequently perform functional, lab-based studies to determine the relationship between genotypes and physiological measures. Examples of measurements used to complement genomic studies include (but are not limited to) $\text{VO}_{2\text{max}}$, forced expiratory volume in one second (FEV1), the proportion of slow-twitch muscle fibres, recovery speed, long-distance running performance, running economy, lactate threshold, erythropoietin, and haemoglobin levels, number of erythrocytes, capillary density, mitochondrial density, fat metabolism, and fatigue resistance. Our literature search revealed that at least 41 of the 115 reported markers are associated with endurance athlete status based on our criteria (Table 4). The most promising genetic markers for endurance athlete status include *AMPD1* rs17602729C, *CDKN1A* rs236448 A, *HFE* rs1799945 G, *MYBPC3* rs1052373 G, *NFIA-AS2* rs1572312C, *PPARA* rs4253778 G, and *PPARGC1A* rs8192678 G alleles. In contrast, the other 74 markers (endurance alleles) have not passed our strict criteria: *ACOXL* rs13027870 G, *ADRA2A* 6.7 kb, *ADRA2A* rs1800544 G, *ADRB1* rs1801252 G, *AGT* rs699 A, *BDKRB2* rs1799722 T, *CAMK1D* rs11257754 A, *CHRNB3* rs4950 G, *CLSTN2* rs2194938 A, *CNDP2* rs6566810 A, *COL5A1* rs17146744 AGGG, *COL6A1* rs35796750 T, *CPQ* rs6468527 A, *CYP2D6* rs3892097 G, *DMT1* 258 bp, *EPAS1* (*HIF2A*) rs1867785 G,

Table 4 Genetic markers for endurance athlete status.

Gene	Full name	Locus	Polymorphism	Endurance-related allele	Studies with positive results	References
ACE	Angiotensin I converting enzyme	17q23.3	Alu I/D or rs4343 A/G or rs4341C/G	I (A or C)	Montgomery et al. (1998), Myerson et al. (1999), Gayagay et al. (1998), Jelaković et al. (2000), Ahmetov et al. (2008d), Alvarez et al. (2000), Collins et al. (2004), Lucia et al. (2005b), Hruskovicová et al. (2006), Nazarov et al. (2001), Scanavini, Bernardi, Castoldi, Conconi, and Mazzoni (2002), Turgut, Turgut, Genc, Atalay, and Atalay (2004), Tsianos et al. (2004), Cieszyżyk, Krupecki, Maciejewska, and Sawczuk (2009), Min et al. (2009), Shenoy, Tandon, Sandhu, and Bhanwer (2010), and Znazen et al. (2016)	Ash et al. (2011), Tobina et al. (2010), Ahmetov et al. (2009b), Papadimitriou, Papadopoulos, Kouvatzi, and Triantaphyllidis (2009), Scott et al. (2005), Rankinen et al. (2000), Taylor, Mamotte, Fallon, and van Bockxmeer (1999), Orysiak et al. (2013), Ginevičienė, Pranckevičienė, Milašius, and Kučinskas (2010), Muniesa et al. (2010), Lucia et al. (2005b), Papadimitriou et al. (2018), and Varillas-Delgado et al. (2022b)

<i>ACTN3</i>	Actinin alpha 3 11q13.1	rs1815739C/T	T	Yang et al. (2003), Shang, Huang, Chang, Zhang, and Huang (2010), Orysiak et al. (2015), and Gasser, Flück, Frey, Valdivieso, and Spörrli (2022) Ahmetov et al. (2010a), Döring et al. (2010a), Ginevičienė et al. (2010), Tsianos et al. (2010), Niemi and Majamaa (2005), Papadimitriou, Papadopoulos, Kouvatsi, and Triantaphyllidis (2008), Paparini et al. (2007), Saunders et al. (2007), Yang et al. (2007), Lucia et al. (2006), Grealy et al. (2013), Mikami et al. (2014), Wang et al. (2013a), Papadimitriou et al. (2018), and Yang et al. (2021)
<i>ADRB2</i>	Adrenoceptor beta 2 5q31-q32	rs1042713 G/A	A	Wagoner et al. (2000), Wolfarth et al. (2007), and Tsianos et al. (2010) Santiago et al. (2011) and Sawczuk, Maciejewska-Karlowska, Gieszczyk, Skotarczak, and Ficek (2013a)

(continued)

Table 4 Genetic markers for endurance athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Endurance-related allele	References	
					Studies with positive results	Studies with negative or controversial results
<i>ADRB2</i>	Adrenoceptor beta 2	5q31-q32	rs1042714 G/C	C	Moore et al. (2001) and Varillas-Delgado et al. (2022b)	Santiago et al. (2011) and Sawczuk et al. (2013a)
<i>ADRB3</i>	Adrenoceptor beta 3	8p11.23	rs4994 A/G	G	Santiago et al. (2011) and Yang et al. (2021)	
<i>AGTR2</i>	Angiotensin II receptor type 2	Xq22-q23	rs11091046 A/C	C	Mustafina et al. (2014)	Guilherme, Silva, Bertuzzi, and Lancha Junior (2018b)
<i>AQP1</i>	Aquaporin 1	7p14	rs1049305C/G	C	Martínez et al. (2009b), Rivera, Martínez, Carrion, and Fahey (2011), and Saunders, Posthumus, O'Connell, September, and Collins (2015)	
<i>AMPD1</i>	Adenosine monophosphate deaminase 1	1p13	rs17602729C/T	C	Rico-Sanz et al. (2003), Thomas et al. (2011), Rubio et al. (2005), Ciężczyk et al. (2011c), and Varillas-Delgado et al. (2022b)	Ginevičienė et al. (2014)

<i>BDKRB2</i>	Bradykinin receptor B2	14q32.1-q32.2	+9/−9 (exon 1)	−9	Williams et al. (2004) and Saunders et al. (2006)	Sawczuk et al. (2013b), Grenda, Leofska-Duniec, Ciężczyk, and Zmijewski (2014), Zmijewski et al. (2016), and Varillas-Delgado et al. (2022b)
<i>CDKN1A</i>	Cyclin Dependent Kinase Inhibitor 1A	6p21.2	rs236448 A/C	A	Semenova et al. (2022)	
<i>CKM</i>	Creatine kinase M-type	19q13.32	rs8111989 A/G	A	Rivera et al. (1997a), Rivera et al. (1999), and Fedotovskaia, Popov, Vinogradova, and Akhmetov (2012)	Rivera et al. (1997b), Lucia et al. (2005b), and Martínez et al. (2009a)
<i>COL5A1</i>	Collagen type V alpha 1 chain	9q34.2-q34.3	rs12722C/T	T	Posthumus, Schwellnus, and Collins (2011) and Brown, Miller, Posthumus, Schwellnus, and Collins (2011)	
<i>FTO</i>	FTO Alpha-Ketoglutarate Dependent Dioxygenase	16q12.2	rs9939609 T/A	T	Guilherme et al. (2019) and Zmijewski and Leofska-Duniec (2021)	Eynon et al. (2013a)

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Table 4 Genetic markers for endurance athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Endurance-related allele	References	
					Studies with positive results	Studies with negative or controversial results
<i>GABPB1</i>	GA binding protein transcription factor subunit beta 1	15q21.2	rs12594956 A/C	A	He et al. (2007) and Eynon et al. (2013b)	
			rs7181866 A/G	G	He et al. (2007) and Maciejewska-Karlowska et al. (2012)	Eynon et al. (2013b)
<i>GALNTL6</i>	Polypeptide N-acetylglucosaminyltransferase 6	4q34.1	rs558129 T/C	C	Rankinen et al. (2016)	
<i>GSTP1</i>	Glutathione S-transferase Pi 1	11q13.2	rs1695 A/G	G	Zarebska et al. (2014b, 2017)	
<i>HFE</i>	Homeostatic iron regulator	6p21.3	rs1799945C/G	G	Deugnier et al. (2002), Chicharro et al. (2004), Hermine et al. (2015), Semenova et al. (2020), and Varillas-Delgado et al. (2022b)	

<i>HIF1A</i>	Hypoxia inducible factor 1 subunit alpha	14q23.2	rs11549465C/T	C	Prior et al. (2003) and Döring et al. (2010b)	Ahmetov et al. (2009b) and Bosnyák et al. (2020)
<i>MCT1</i>	Monocarboxylate transporter 1	1p12	rs1049434 A/T	T	Merzhinskaya, Fishbein, Davis, and Foellmer (2000), Cupeiro, Benito, Maffulli, Calderón, and González-Lamuño (2010), Fedotovskaya, Mustafina, Popov, Vinogradova, and Ahmetov (2014), Guilherme et al. (2021a), and Ramírez de la Piscina-Viúdez et al. (2021)	Sawczuk et al. (2015)
MtDNA loci	Mitochondrial DNA	MtDNA	MtDNA haplogroups	H	Niemi and Majamaa (2005) and Maruszak et al. (2014)	
<i>MYBPC3</i>	Myosin Binding Protein C3	11p11.2	rs1052373 A/G	G	Unfavourable: K	Niemi and Majamaa (2005) and Maruszak et al. (2014)
					Al-Khelaifi et al. (2020)	

(continued)

Table 4 Genetic markers for endurance athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Endurance-related allele	References	
					Studies with positive results	Studies with negative or controversial results
<i>NFATC4</i>	Nuclear factor of activated T cells 4	14q11.2	rs2229309 G/C	G	Ahmetov et al. (2009b)	
<i>NFIA-AS2</i>	NFIA antisense RNA 2	1p31.3	rs1572312C/A	C	Ahmetov et al. (2015) , and Malczewska-Lenczowska et al. (2022)	
<i>NOS3</i>	Nitric oxide synthase 3	7q36	rs2070744 T/C	T	Drozdowska, Dosenko, Ilyin, Filippov, and Kuzmina (2009) , Zmijewski et al. (2018) , and Varillas-Delgado et al. (2022b)	Gómez-Gallego et al. (2009a)
<i>PPARA</i>	Peroxisome proliferator activated receptor alpha	22q13.31	rs4253778 G/C	G	Ahmetov et al. (2006) , Akhetov et al. (2007b) , Maciejewska, Sawczuk, and Cięszczyk (2011) , and Tural et al. (2014)	

<i>PPARGC1A</i>	Peroxisome proliferative activated receptor, gamma, coactivator 1 alpha	4p15.1	rs8192678	G/A	G	Lucia et al. (2005a), Akhmetov et al. (2007b), Maciejewska, Sawczuk, Cieszczyk, Mozhayskaya, and Akhmetov (2012), and Yang et al. (2021) Maruszak et al. (2014), He et al. (2015), and Hall et al. (2023)
<i>PPARGC1B</i>	Peroxisome proliferative activated receptor, gamma, coactivator 1 beta	5q32	rs7732671	G/C	C	Akhmetov et al. (2009b), and Akhmetov, Popov, Missina, Vinogradova, and Rogozkin (2009)
<i>PPP3R1</i>	Protein phosphatase 3 regulatory subunit B, alpha	2p15	Promoter	5I/5D	5I	Akhmetov et al. (2009b), and Akhmetov et al. (2008)
<i>PRDM1</i>	PR/SET Domain 1	6q21	rs10499043C/T	T	T	Bouchard et al. (2011), and Hall et al. (2021a)

(continued)

Table 4 Genetic markers for endurance athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Endurance-related allele	References	
					Studies with positive results	Studies with negative or controversial results
<i>RBFOX1</i>	RNA binding fox-1 homolog 1	16p13.3	rs7191721 G/A	G	Ahmetov et al. (2015)	
<i>SIRT1</i>	Sirtuin 1	10q21.3	rs41299232C/G	G	Bulgay et al. (2023)	
<i>SPEG</i>	Striated Muscle Enriched Protein Kinase	2q35	rs7564856 G/A	G	Kusić et al. (2020)	
<i>TFAM</i>	Transcription factor A, mitochondrial	10q21	rs1937 G/C	C	Ahmetov et al. (2009b) , Ahmetov, Popov, Missina, Vinogradova, and Rogozkin (2010b)	Maruszak et al. (2014)
<i>TRPM2</i>	Transient Receptor Potential Cation Channel Subfamily M Member 2	21q22.3	rs1785440 A/G	G	Bulgay et al. (2023)	

<i>TSHR</i>	Thyroid stimulating hormone receptor	14q31	rs7144481 T/C	C	Ahmetov et al. (2015)
<i>UCP2</i>	Uncoupling protein 2	11q13	rs660339C/T	T	Ahmetov et al. (2008d) , Ahmetov et al. (2009b) , and Gronek et al. (2018)
<i>UCP3</i>	Uncoupling Protein 3	11q13	rs1800849C/T	T	Ahmetov et al. (2008d) and Hudson, Mokone, Noakes, and Collins (2004) Ahmetov et al. (2009b)
<i>VEGFA</i>	Vascular endothelial growth factor A	6p12	rs2010963 G/C	C	Prior et al. (2006) , Ahmetov et al. (2008b) , and Ahmetov et al. (2009b)
<i>VEGFR2</i>	Vascular endothelial growth factor receptor 2	4q11-q12	rs1870377 T/A	A	Ahmetov et al. (2009a) and Eider et al. (2013b)

EPAS1 (HIF2A) rs11689011 T, *GABPB1* rs8031031 T, *GALM* rs3821023 A, *GNB3* rs5443 T, *GRM3* rs724225 G, *GSTT* rs17856199 (+), *IGF1R* rs1464430 A, *IL6* rs1800795C, *IL15RA* rs2228059 A, *ITPR1* rs1038639 T, *ITPR1* rs2131458 T, *FMNL2* rs12693407 G, *KCNJ11* rs5219C, *L3MBTL4* rs17483463 T, *MSTN* rs11333758 D, MtDNA loci (G1, HV, L0, M*, m .11215T, m .152C, m .15518T, m .15874G, m .4343G, m .514(CA)≤ 4, poly(C≥7) stretch at m.568–573, m .16080G, m .5178C, N9, V, unfavourable: B, J2, T, L3*), *NALCN-AS1* rs4772341 A, *NACC2* rs4409473 C, *NATD1* rs732928 G, *NOS3* rs1799983 G, *NOS3* (CA)_n 164 bp, *NOS3* 27 bp 4B, *PPARD* rs2016520C, *PPARD* rs1053049 T, *PPARGC1A* rs4697425 A, *PPARGC1B* rs11959820 A, *PPP3CA* rs3804358C, *PPP3CB* rs3763679C, *RAB3GAP2* rs115660502 G, *SGMS1* rs884880 A, *SLC2A4* rs5418 A, *SOD2* rs4880C, *SPOCK1* rs1051854 T, *TPK1* rs10275875 T, *TTN* rs10497520 T, Y-chromosome haplogroups (E*, E3* and K*(xP); unfavourable: E3b1), and *ZNF429* rs1984771 G. Most of these markers have been described in previous reviews (Ahmetov & Fedotovskaya, 2015; Ahmetov, Egorova, Gabdrakhmanova, & Fedotovskaya, 2016a; Semenova et al., 2019a) but cannot be included in our current list of endurance-associated markers until they are validated through replication by additional studies.

4.2 Genetic profile of power athletes

Several characteristics are positively associated with power performance, including circulating levels of testosterone, proportion of fast-twitch muscle fibres, muscle mass and strength, body and calcaneus height, muscle fascicle length, and reaction time, among others (Ahmetov et al., 2020; Hall et al., 2021b; Haugen, Seiler, Sandbakk, & Tønnessen, 2019; Kumagai et al., 2000; Suga et al., 2020; Tønnessen, Haugen, & Shalfawi, 2013; Uth, 2005; Weyand & Davis, 2005). The heritability of power-related phenotypes has been reported in the literature to range from approximately 49 to 86% in a range of phenotypes, including jumping ability (Hughes, Day, Ahmetov, & Williams, 2011; Zempo et al., 2017). Genetic variants associated with power athlete status are typically determined by comparing allelic frequencies between power athletes (e.g., sprinters, throwers, jumpers, wrestlers, gymnasts, etc.) and non-athletic controls. To support findings from power-related case-control studies, researchers perform functional studies to determine the relationship between genotypes and physiological measures that may include sprint times, jump performance, muscle fibre size, muscle fibre typology, maximal strength, rate of force

development, and circulatory levels of anabolic hormones such as testosterone. Our literature search revealed that at least 45 of the 96 markers reportedly associated with power athlete status met our new criteria (Table 5). The most promising of these genetic markers associated with power athlete status currently include *ACTN3* rs1815739C, *AMPD1* rs17602729C, *CDKN1A* rs236448C, *CPNE5* rs3213537 G, *GALNTL6* rs558129 T, *IGF2* rs680 G, *IGSF3* rs699785 A, *NOS3* rs2070744 T and *TRHR* rs7832552 T alleles. In contrast, the remaining 51 genetic markers (power alleles) did not meet our strict criteria: *ARHGEF28* rs17664695 G, *CACNG1* rs1799938 A, *CALCR* rs17734766 G, *CLSTN2* rs2194938C, *CNDP1* rs2887 A, *CNDP1* rs2346061C, *CNDP2* rs3764509 G, *COTL1* rs7458 T, *CREM* rs1531550 A, *DMD* rs939787 T, *EPAS1* (*HIF2A*) rs1867785 G, *EPAS1* (*HIF2A*) rs11689011C, *FOCAD* rs17759424C, *GABRR1* rs282114 A, *GALNT13* rs10196189 G, *GPC5* rs852918 T, *IGF1R* rs1464430C, *IL1RN* rs2234663*2, *IP6K3* rs6942022C, *MCT1* rs1049434 A, *MED4* rs7337521 T, *MDM4* rs35493922 D, *MPRIIP* rs6502557 A, MtDNA loci (favourable: F, m .204C, m .151T, m .15314A, Non-L/UCięszczyk6, unfavourable: m .16278T, m .5601T, m .4833G, m .5108C, m .7600A, m .9377G, m .13563G, m .14200C, m .14569A), *MTR* rs1805087 G, *MTRR* rs1801394 G, *NOS3* rs1799983 G, *NRG1* rs17721043 A, *PPARGC1A* rs8192678 A, *PPARGC1B* rs10060424C, *RC3H1* rs767053 G, *SLC6A2* rs1805065C, *SUCLA2* rs10397 A, *TPK1* rs10275875C, *UCP2* rs660339C, *VEGFR2* rs1870377 T, *WAPL* rs4934207C, *ZNF423* rs11865138C. The majority of these markers are reported in previous reviews (Ahmetov & Fedotovskaya, 2015; Ahmetov et al., 2016; Maciejewska-Skrendo et al., 2019) and should be validated in additional studies before they can meet the criteria to be included in our list of power-associated genetic variants.

4.3 Genetic profile of strength athletes

Performance in strength-based sports is based on multiple factors. However, the factors considered to contribute substantially to strength phenotypes include skeletal muscle hypertrophy (increased cross-sectional area of muscle fibres), hyperplasia, the predominance of fast-twitch muscle fibres, a greater muscle fascicle pennation angle, improved neurological adaptation, high glycolytic capacity, and increased circulatory testosterone (Kuschel, Sonnenburg, & Engel, 2022). Importantly, evidence exists that strength athletes exhibit vastly different transcriptomic, biochemical, anthropometric, physiological, and biomechanical characteristics compared

Table 5 Genetic markers for power athlete status.

Gene	Full name	Locus	Polymorphism	Power-related allele	Studies with positive results	References
ACE	Angiotensin I converting enzyme	17q23.3	Alu I/D or rs4343 A/Gor rs4341C/G	D (G)	Myerson et al. (1999), Nazarov et al. (2001), Woods et al. (2001), Papadimitriou et al. (2009), Costa et al. (2009a), Boraïta et al. (2010), Wang et al. (2013a), and Papadimitriou et al. (2016)	Amir et al. (2007), Kim et al. (2010), Ginevičienė, Pranculis, Jakaitienė, Milašius, and Kučinskas (2011), Shahmoradi, Ahmadalipour, and Salehi (2014), and Scott et al. (2010)
ACVR1B	Activin A type IB receptor	12q13.13	rs2854464 A/G	A	Windelinckx et al. (2011) and Voisin et al. (2016)	Voisin et al. (2016) and Venckunas and Degens (2022)

<i>ACTN3</i>	Actinin alpha 3	11q13.1	rs1815739C/T	C	Yang et al. (2003), Niemi and Majamaa (2005), Papadimitriou et al. (2008), Druzhevskaya, Ahmetov, Astratenkova, and Rogozkin (2008), Massidda, Vona, and Calò (2009), Chiu et al. (2011), Ahmetov et al. (2011), Ciężczyk et al. (2011a), Ciężczyk et al. (2011b), Mikami et al. (2014), Papadimitriou et al. (2016), Yang et al. (2017), Ben-Zaken, Eliakim, Nemet, and Meckel (2019), Yang et al. (2021), and Akazawa et al. (2022) Ginevičienė et al. (2010), Scott et al. (2010), Yang et al. (2007), Koku et al. (2019), and Zaccagni et al. (2019)
<i>ADAM15</i>	ADAM Metallopeptidase Domain 15	1q21.3	rs11264302 G/A	G	Guilherme et al. (2022c)

(continued)

Table 5 Genetic markers for power athlete status. (cont'd)

Gene	Full name	Locus	Polymorphism	Power-related allele	Studies with positive results	References
<i>ADRB2</i>	Adrenoceptor beta 2	5q31-q32	rs1042713 G/A	G	Sawczuk et al. (2013a) and Pickering et al. (2019)	
				G	Sawczuk et al. (2013a) and Pickering et al. (2019)	
<i>AGRN</i>	Agrin	1p36.33	rs4074992C/T	C	Bulgay et al. (2023)	
<i>AGT</i>	Angiotensinogen	1q42.2	rs699 T/C	C	Gomez-Gallego et al. (2009b), Zarębska et al. (2013), and Pickering et al. (2019)	
<i>AGTR2</i>	Angiotensin II receptor type 2	Xq22-q23	rs11091046 A/C	A	Mustafina et al. (2014) and Guilherme et al. (2018b)	Yvert et al. (2018)
<i>AKAP6</i>	A-Kinase Anchoring Protein 6	14q12	rs12883788C/T	C	Guilherme et al. (2022c)	

<i>AMPD1</i>	Adenosine monophosphate deaminase 1	1p13	rs17602729C/T	C	Cieszczyk et al. (2012), Fedotovskaya, Danilova, and Akhmetov (2013a), and Ginevičienė et al. (2014)
<i>AUTS2</i>	Activator of Transcription and Developmental Regulator AUTS2	7q11.22	rs10452738 A/G	A	Guilherme et al. (2022c)
<i>BDNF</i>	Brain derived neurotrophic factor	11p14.1	rs10501089 G/A	A	Guilherme et al. (2022a)
<i>CCT3</i>	Chaperonin Containing TCP1 Subunit 3	1q22	rs11548200 T/C	T	Guilherme et al. (2022c)
<i>CDKN1A</i>	Cyclin Dependent Kinase Inhibitor 1A	6p21.2	rs236448 A/C	C	Semenova et al. (2022)

(continued)

Table 5 Genetic markers for power athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Power-related allele	Studies with positive results	References
<i>CKM</i>	Creatinekinase, M-type	19q13.32	rs8111989 A/G	G	Fedotovskaya et al. (2013b), He, Li, Qian, and Yan (2016), and Saito et al. (2022b)	Ginevičiūnė et al. (2021)
<i>CNTFR</i>	Ciliary neurotrophic factor receptor	9p13.3	rs41274853C/T	T	Miyamoto-Mikami et al. (2016)	
<i>CPNE5</i>	Copine V	6p21.2	rs3213537 G/A	G	Pickering et al. (2019) and Guilherme et al. (2021b)	
<i>CRTAC1</i>	Cartilage Acidic Protein 1	10q24.2	rs2439823 A/G	A	Guilherme et al. (2022c)	
<i>CRTC1</i>	CREB Regulated Transcription Coactivator 1	19p13.11	rs11881338 T/A	A	Guilherme et al. (2022c)	

<i>E2F3</i>	E2F Transcription Factor 3	6p22.3	rs4134943C/T	T	Guilherme et al. (2022c)
<i>FHL2</i>	Four and a Half LIM Domains 2	2q12.2	rs55680124C/T	C	Guilherme et al. (2022c)
<i>GALNTL6</i>	Polypeptide N-acetylga- lactosaminyl- transferase like 6	4q34.1	rs558129C/T	T	Díaz Ramírez et al. (2020) and Zmijewski, Trybek, Czarny, and Leońska-Duniec (2021)
<i>GDF5</i>	Growth Differentiation Factor 5	20q11.22	rs143384 A/G	G	Guilherme et al. (2022c)
<i>HIF1A</i>	Hypoxia inducible factor 1 alpha subunit	14q21-q24	rs11549465C/T	T	Cięszczyk et al. (2011a), Gabbasov et al. (2013), and Drozdowska, Dosenko, Ahmetov, and Ilyin (2013)

(continued)

Table 5 Genetic markers for power athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Power-related allele	Studies with positive results	References
<i>HSD17B14</i>	Hydroxysteroid 17- β dehydrogenase 14	19q13.33	rs7247312 A/G	G	Pickering et al. (2019)	
<i>IGF1</i>	Insulin like growth factor 1	12q23.2	rs35767C/T	T	Ben-Zaken, Meckel, Nemet, and Eliakim (2013) and Ben-Zaken et al. (2022)	
<i>IGF2</i>	Insulin like growth factor 2	11p15.5	rs680 A/G	G	Itaka, Agemizu, Aruga, and Machida (2016), Pickering et al. (2019), and Ben-Zaken et al. (2022)	
<i>IGSF3</i>	Immunoglobulin Superfamily Member 3	1p13.1	rs699785 G/A	A	Guilherme et al. (2022c)	

<i>IL6</i>	Interleukin 6	7p21	rs1800795C/G	G	Ruiz et al. (2010), Eider et al. (2013a), and Pickering et al. (2019)
<i>ILRN</i>	Inflammation and Lipid Regulator with UBA-Like and NBR1-Like Domains	6p21.31	rs205262 A/G	A	Guilherme et al. (2022c)
<i>MTHFR</i>	Methylenetetrahydrofolate reductase	1p36.3	rs1801131 A/C	C	Terruzzi et al. (2011) and Zarebska et al. (2014a)
<i>NOS3</i>	Nitric oxide synthase 3	7q36	rs2070744 T/C	T	Drozdzowska et al. (2013), Gómez-Gallego et al. (2009a), Gomez-Gallego et al. (2009b), and Murtagh et al. (2020)
<i>NRXN3</i>	Neurexin 3	14q24.3-q31.1	rs8011870 G/A	G	Guilherme et al. (2022c)

(continued)

Table 5 Genetic markers for power athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Power-related allele	References	
					Studies with positive results	Studies with negative or controversial results
<i>NUP210</i>	Nucleoporin 210	3p25.1	rs2280084C/A	C	Bulgay et al. (2023)	
<i>PIEZO1</i>	Piezo Type Mechanosensitive Ion Channel Component 1	16q24.3	rs572934641 (TCC/-) E756del	D	Nakamichi et al. (2022)	
<i>PPARA</i>	Peroxisome proliferator activated receptor alpha	22q13.31	rs4253778 G/C	C	Ahmetov et al. (2006) , Ginevičienė et al. (2010) , and Maciejewska-Skrendo et al. (2021)	

<i>PPARG</i>	Peroxisome proliferator activated receptor gamma	3p25.2	rs1801282C/G	G	Ahmetov, Mozhayskaya, Lyubaeva, Vinogradova, and Rogozkin (2008c), Maciejewska-Karlowska, Sawczuk, Cieszyk, Zarebska, and Sawczyn (2013), and Drozdovska et al. (2013)	Guilherme, Bertuzzi, Lima-Silva, Pereira, and Lancha Junior (2018a)
					rs2920503C/T	T
<i>SLC39A8</i>	Solute Carrier Family 39 Member 8	4q24	rs13107325C/T	C	Guilherme et al. (2022c)	Guilherme et al. (2022c)
<i>SOD2</i>	Superoxide dismutase 2	6q25.3	rs4880C/T	C	Ahmetov et al. (2014b)	
<i>TRHR</i>	Thyrotropin releasing hormone receptor	8q23.1	rs7832552C/T	T	Miyamoto-Mikami et al. (2017), Khanal et al. (2020), and Saito et al. (2022b)	

(continued)

Table 5 Genetic markers for power athlete status. (cont'd)

Gene	Full name	Locus	Polymorphism	Power-related allele	Studies with positive results	References
UBR5	Ubiquitin Protein Ligase E3 Component N-Recognin 5	8q22.3	rs10505025 G/A	A	Seaborne et al. (2019)	Studies with negative or controversial results
			rs47344621 G/A	A	Seaborne et al. (2019)	
ZNF568	Zinc Finger Protein 568	19q13.12	rs1667369 A/C	A	Guilherme et al. (2022c)	

to endurance athletes and/or non-athletic cohorts (Fuku, Kumagai, & Ahmetov, 2019; Stepto et al., 2009). These differences can be explained by deliberate environmental (training, nutrition, etc.) and genetic factors. Indeed, studies indicate a strong heritability of power and strength-related traits, where genetic factors account for 30–85% of the variation in maximal isometric, isotonic, and isokinetic strength (Zempo et al., 2017). In a recent study investigating the genetic component of severe sarcopenia (the age-related decline in skeletal muscle mass, strength, and gait speed) (Semenova et al., 2023b), it was found that the alleles associated with a higher risk of severe sarcopenia were closely linked to tiredness, reported frequency of falls in the last year, neuroticism, frequency of alcohol intake, smoking, time spent watching television, and higher self-reported consumption of salt, white bread, and processed meat.

In contrast, alleles associated with lower risk of severe sarcopenia were positively associated with bone mineral density, levels of serum testosterone, IGF1, and 25-hydroxyvitamin D, height, intelligence, cognitive performance, educational attainment, income, physical activity, ground coffee drinking, as well as indicators of healthier dietary habits (self-reported intake of muesli, cereal, wholemeal or wholegrain bread, potassium, magnesium, cheese, oily fish, protein, water, fruit and vegetables). Whilst muscle strength phenotypes in the general population may be less pronounced than in strength athletes, the latter may represent an ideal population to identify genomic variants associated with skeletal muscle capacity, potentially aiding the advancement of knowledge surrounding sarcopenia and directing strategies to reduce the negative impact of age-related declines in muscle mass. Genetic variants associated with strength athlete status can be determined by comparing allelic frequencies between strength athletes (e.g., weightlifters, powerlifters etc.) and non-athletic controls. To support findings from strength-related case-control studies, researchers perform functional studies to quantify relationships between genotypes and physiological measures, including maximal strength as measured by handgrip dynamometry, isokinetic dynamometry, powerlifting/weightlifting performance, as well as measuring the acute and chronic responses to resistance training.

Handgrip strength is one of the most studied traits in relation to sports and exercise genomics. Using a GWAS approach, 170 DNA polymorphisms were associated with handgrip strength in three large studies (Jones et al., 2021; Tikkanen et al., 2018; Willems et al., 2017). In a subsequent study involving elite strength athletes, Moreland et al. (2022) tested the

Table 6 Genetic markers for strength athlete status.

Gene	Full name	Locus	Polymorphism	Strength-related allele	References	
					Studies with positive results	Studies with negative or controversial results
<i>ABHD17C</i>	Abhydrolase domain containing 17C	15q25.1	rs7165759 G/A	A	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>ACE</i>	Angiotensin I converting enzyme	17q23.3	Alu I/Dor rs4343 A/Gor rs4341C/G	D (G)	Colakoglu et al. (2005) , Giaccaglia et al. (2008) , Costa et al. (2009b) , Pimjan et al. (2017) , Melían Ortiz, Laguarda-Val, and Varillas-Delgado (2021) , and Khanal et al. (2022)	Gineviciene et al. (2016)
<i>ACTG1</i>	Actin gamma 1	17q25.3	rs6565586 T/A	A	Willems et al. (2017) and Moreland et al. (2022)	

<i>ACTN3</i>	Actinin alpha 3	11q13.1	rs1815739C/T	C	Roth et al. (2008), Erskine, Williams, Jones, Stewart, and Degens (2014), Pimjan et al. (2017), and Homma et al. (2022)	Gineviciene et al. (2016), Orysiak et al. (2018), Ben-Zaken et al. (2019), and Moreland et al. (2022)
<i>ADCY3</i>	Adenylate cyclase 3	2p23.3	rs10203386 T/A	T	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>ADPGK</i>	ADP dependent glucokinase	15q24.1	rs4776614C/G	C	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>AGT</i>	Angiotensinog- en	1q42.2	rs699 T/C	C	Aleksandra et al. (2016) and Ben-Zaken et al. (2019)	Moreland et al. (2022)
<i>ALDH2</i>	Aldehyde Dehydrogenase 2 Family Member	12q24.12	rs671 G/A	G	Kikuchi et al. (2022b), De Almeida et al. (2022b), and Saito et al. (2022a)	

(continued)

Table 6 Genetic markers for strength athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Strength-related allele	References	
					Studies with positive results	Studies with negative or controversial results
<i>ANGPT2</i>	Angiopoietin 2	8p23.1	rs890022 G/A	A	Matteini et al. (2016) and Moreland et al. (2022)	
<i>AR</i>	Androgen Receptor	Xq12	(CAG) _n	≥ 21	Walsh et al. (2005) and Guilherme et al. (2021c)	
<i>ARPP21</i>	CAMP regulated phosphoprotein 21	3p22.3	rs1513475 T/C	C	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>BCDIN3D</i>	Bicoid interacting 3 domain containing RNA methyltransferase	12q13.12	rs12367809C/T	C	Tikkanen et al. (2018) and Moreland et al. (2022)	

<i>CKM</i>	Creatine kinase, M-type	19q13.32	rs8111989	A/G	G	Fedotovskaia et al. (2012) and Chen, Sun, Liang, Yu, and Hu (2017)	Moreland et al. (2022) and Ginevičienė et al. (2021)
<i>CNTFR</i>	Ciliary neurotrophic factor receptor	9p13.3	rs41274853C/T	T	T	Miyamoto-Mikami et al. (2016) and Homma et al. (2020)	Moreland et al. (2022)
<i>CRTAC1</i>	Cartilage acidic protein 1	10q24.2	rs563296	G/A	G	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>DHODH</i>	Dihydroorotate dehydrogenase (Quinone)	16q22.2	rs12599952	G/A	A	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>GALNTL6</i>	Polypeptide N-acetylgalactosaminyltransferase like 6	4q34.1	rs558129C/T	T	T	Díaz Ramírez et al. (2020)	
<i>GBE1</i>	1, 4-Alpha-glucan branching enzyme 1	3p12.2	rs9877408	A/G	A	Tikkanen et al. (2018) and Moreland et al. (2022)	

(continued)

Table 6 Genetic markers for strength athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Strength-related allele	References	
GBF1	Golgi brefeldin A resistant guanine nucleotide exchange factor 1	10q24.32	rs2273555 G/A	A	Studies with positive results	Studies with negative or controversial results
					Willem's et al. (2017) and Grishina et al. (2019)	
GLIS3	GLIS Family Zinc Finger 3	9p24.2	rs34706136 T/TG	TG	Guilherme et al. (2022b)	
HIF1A	Hypoxia inducible factor 1 alpha	14q21-q24	rs11549465C/T	T	Ahmetov, Hakimullina, Lyubaeva, Vinogradova, and Rogozkin (2008a), Gabbasov et al. (2013), and Khanal et al. (2020)	Moreland et al. (2022)
IGF1	Insulin like growth factor 1	12q23.2	rs35767C/T	T	Kostek et al. (2010), Ben-Zaken et al. (2013), and Moreland et al. (2022)	

<i>IL6</i>	Interleukin 6	7p21	rs1800795C/G	G	Ruiz et al. (2010) and Moreland et al. (2022)
<i>ITPR1</i>	Inositol 1, 4, 5-Triphosphate Receptor Type 1	3p26.1	rs901850 G/T	T	Tikkanen et al. (2018) and Moreland et al. (2022)
<i>KIF1B</i>	Kinesin family member 1B	1p36.22	rs11121542 G/A	G	Tikkanen et al. (2018) and Moreland et al. (2022)
<i>LRPPRC</i>	Leucine-rich pentatricopeptide repeat cassette	2p21	rs10186876 A/G	A	Willem's et al. (2017) , Moreland et al. (2022) , and Kikuchi et al. (2022a)
<i>MLN</i>	Motilin	6p21.31	rs12055409 A/G	G	Tikkanen et al. (2018) and Grishina et al. (2019)
<i>MMS22L</i>	Methyl methanesulfonyl-sensitivity protein 22-Like	6q16.1	rs9320823 T/C	T	Tikkanen et al. (2018) , Moreland et al. (2022) , and Kikuchi et al. (2022a)

(continued)

Table 6 Genetic markers for strength athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Strength-related allele	References	
					Studies with positive results	Studies with negative or controversial results
<i>MTHFR</i>	Methylenetetrahydrofolate reductase	1p36.3	rs1801131 A/C	C	Zarębska et al. (2013) , Moreland et al. (2022) , and Kikuchi et al. (2022a)	
<i>NPIP6</i>	Nuclear pore complex interacting protein family member B6	16p12.1	rs2726036 A/C	A	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>PHACTR1</i>	Phosphate and actin regulator 1	6p24.1	rs6905419C/T	C	Tikkanen et al. (2018) , Moreland et al. (2022) , and Kikuchi et al. (2022a)	
<i>PLEKHB1</i>	Pleckstrin homology domain containing B1	11q13.4	rs7128512 A/G	G	Matteini et al. (2016) and Moreland et al. (2022)	

<i>PPARA</i>	Peroxisome proliferator activated receptor alpha	22q13.31	rs4253778 G/C	C	Ahmetov et al. (2006), Ahmetov et al. (2013), and Petr et al. (2014)	Moreland et al. (2022)
<i>PPARG</i>	Peroxisome proliferator activated receptor gamma	3p25.2	rs1801282C/G	G	Ahmetov et al. (2008c), Maciejewska-Karlowska et al. (2013), and Moreland et al. (2022)	
<i>PPARGC1A</i>	Peroxisome proliferative activated receptor, gamma, coactivator 1 alpha	4p15.2	rs8192678 G/A	A	Gineviciene et al. (2016), Moreland et al. (2022), and Homma et al. (2022)	
<i>R3HDM1</i>	R3H domain containing 1	2q21.3	rs6759321 G/T	T	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>RASGRF1</i>	Ras protein specific guanine nucleotide Releasing Factor 1	15q25.1	rs1521624C/A	A	Tikkanen et al. (2018) and Moreland et al. (2022)	

(continued)

Table 6 Genetic markers for strength athlete status. (*cont'd*)

Gene	Full name	Locus	Polymorphism	Strength-related allele	References	
					Studies with positive results	Studies with negative or controversial results
<i>RMCI</i>	Regulator of MON1-CCZ1	18q11.2	rs303760C/T	C	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>SLC39A8</i>	Solute carrier family 39 member 8	4q24	rs13135092 A/G	A	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>TFAP2D</i>	Transcriptional factor AP-2 delta	6p12.3	rs56068671 G/T	T	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>ZKSCAN5</i>	Zinc finger with KRAB and SCAN domains 5	7q22.1	rs3843540 T/C	C	Tikkanen et al. (2018) and Moreland et al. (2022)	
<i>ZNF608</i>	Zinc finger protein 608	5q23.2	rs4626333 G/A	G	Matteini et al. (2016) and Grishina et al. (2019)	

hypothesis that alleles associated with greater handgrip strength would be over-represented in weightlifters and powerlifters compared to controls. Indeed, they found that 23 SNPs were associated with strength athlete status. Interestingly, of these 23 SNPs, the *LRPPRC* rs10186876, *MMS22L* rs9320823, and *PHACTR1* rs6905419 polymorphisms were also associated with superior competitive weightlifting performance (total lifts in snatch and clean and jerk, adjusted for sex and weight) (Kikuchi et al., 2022a).

Our literature search based on our new inclusion criteria revealed that at least 42 genetic markers are associated with strength athlete status (Table 6). The most promising genetic markers for strength athlete status include *ACTN3* rs1815739C, *AR* ≥ 21 CAG repeats, *LRPPRC* rs10186876 A, *MMS22L* rs9320823 T, *PHACTR1* rs6905419C, and *PPARG* rs1801282 G alleles.



5. Conclusions

The current review demonstrates that at least 149 variants have been associated with various physical activity traits, and 253 genetic markers are reportedly linked to sport-related traits. As discussed previously, height remains not only the most studied exercise-related phenotype at the genetic level but also the most studied human trait, with 12,111 associated SNPs (Yengo et al., 2022). It is estimated that the final number of height-related SNPs may reach 25,000 (with a minor allele frequency of $\geq 1\%$), but this will require an increase in sample size to approximately 100 million individuals of the same ethnicity. These values should be noteworthy and serve as a benchmark for the direction of future research in the field of genomics of physical activity, exercise, and sport, where the current number of genetic markers must be increased by a considerable magnitude to fully comprehend the genomic underpinnings of exercise performance and thus to be considered as potential predictors of both physical activity and sporting talent.

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