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Physical Activity, Physical Fitness, Gross Motor Coordination, and Metabolic Syndrome: Focus of Twin Research in Portugal

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A very brief history of Portuguese twin research in sport and human movement sciences is presented. Recruitment procedures, zygosity determination, and phenotypes are given for twins and their parents from the mainland, and Azores and Madeira archipelagos. Preliminary findings are mostly related to physical activity, health-related physical fitness, gross motor coordination, neuromotor development, and metabolic syndrome traits.

Keywords: Portugal, twins, physical activity, physical fitness, gross motor coordination, metabolic syndrome

Brief History

The Portuguese territory comprises a mainland located in the most western part of Europe, and two archipelagos both in the Atlantic Ocean: Azores (nine islands), and Madeira (two islands). The Portuguese twin research started in 1999 in the mainland by the first author (JARM), and then expanded to the Azores in 2001. In 2004, together with DLF, a new research branch started in Madeira. The focus of our research is on human physical activity and related traits, including physical growth, biological maturation, health perceptions, physical fitness, gross motor coordination, neuromotor development, body composition, bone, and metabolic syndrome (MetS). Our research started without funding. It was only in 2002 that the Portuguese Foundation for Science (FCT) gave a grant to JARM to study twins' physical activity, physical fitness, and body composition. Later on, in 2004, another grant from FCT allowed a joint venture with DLF, the late Gaston Beunen and Martine Thomis from the Katholieke Universiteit Leuven, Belgium, and colleagues from the University of Madeira Genetics Department. This 'research adventure', named genetic and environmental influences in physical activity, physical fitness and health (GEAFAS) (Madeira Family Study), was broad in its sampling schema (three-generation families) and scope (from quantitative to molecular genetics probing the value of several candidate genes). As no published twin research was available in the Portuguese language, we produced several papers regarding twin methodology and books/reports for Portuguese-speaking sport and human movement sciences researchers from Portugal, Brazil, Mozambique, and Angola (Maia, 2001; Maia et al., 2004, 2011; Sapage & Maia, 2007). Furthermore, over the years, and in conjunction with the Portuguese Twin Group Register, four twin meetings were organized around the theme of active and healthy lifestyles.

Recruitment Procedures

As the main focus of our twin research is physical activity and related traits during the childhood and adolescent growth period, our target twin population was schoolaged children aged 4–18 years. Twin recruitment was made in several ways: first, through school lists and information from physical education teachers; second, through announcements in local newspapers and TV; third, through advertisements with local governmental bodies that facilitated all our twin meetings; fourth, through the Portuguese Twin Group.

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TABLE 1	
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Domains (test batteries)	Phenotypes	Equipment
Human physical growth (International Biological Program) Biological maturation Body composition	Height, weight, skinfolds, breaths, circumferences, lengths Bone age (Tanner-Whitehouse III) % body fat, lean body mass, % trunk, arms and legs fat and mass	Harpenden anthropometer, skinfold calipers, and Seca scale Portable x-ray machine Tanita BC-418 MA
Bone accrual/health Physical activity	Bone mineral content and density Physical activity at work/school, leisure time, sport, total; Counts and METs (different intensity levels); Steps and calories; Approximate entropy patterns	DXA Questionnaires (Baecke et al., 1982; Godin & Shephard, 1985); accelerometer (TRITRAC R3D); pedometer (Yamax DW-SW 700)
Physical fitness (Fitnessgram test battery)	1-mile run/walk, curl-ups, push-ups, trunk lift	Standard equipment provided by manufacturer
Gross motor coordination (Körperkoordinationtest für Kinder: KTK)	Backward balance (BB), hoping in one leg (HP), shifting platforms (SP), jumping sideways (JS); global motor coordination quotient (MCC)	Standard equipment provided by manufacturer
Neuromotor development (Zurich neuromotor assessment)	Purely motor tasks, fine motor task adaptive, dynamic balance, and associated movements	Standard equipment provided by manufacturer; video camera Sony, 50 Hz
Health-related quality of life	Physical functioning, Role-physical, Bodily pain, General health, Vitality, Social functioning, Role-emotional, Mental health	SF-36 Questionnaire
Physical self-concept	Strength, body fat, physical activity, endurance/fitness, sports competence, coordination, appearance, flexibility, health	Physical Self-Description Questionnaire
Metabolic syndrome	Glucose, triglycerides, HDL-cholesterol, blood pressure, waist girth	OMRON – M10 IT; Cholestech LDX analyzers

Domains, Test Batteries, Phenotypes, and Equipment

Zygosity Assignment

Two procedures were used to assess zygosity: (1) by the Peeters et al. (1998) questionnaire, that was later crossvalidated in Portugal using DNA data (Maia et al., 2007); (2) by DNA extraction with a method based on the use of Chelex resin. Genotyping was performed on an ABI 310 Genetic Analyzer (AB Applied Biosystems, Carlsbad, California, USA), according to the manufacturer's instructions. In all DNA samples, the analysis of 17 short tandem repeats autosomal (CSF1PO, D2S1338, D3S1358, D5S818, D7S820, D8S1179, D13S317, D16S539, D18S51, D19S433, D21S11, FGA, PD, PE, TH01, TPO, and VWA) and the Amelogenin locus (sex determination) was performed. Allele frequencies of the different genetic markers in the north and center of Portugal were used in the computation of probabilities of being monozygotic (MZ) twins.

Measures

Table 1 presents research domains, tests used, phenotypes, and equipment used in different twin studies. Twin research in Portugal has been initiated from scientists within the field of sport and movement sciences, with a special focus on children and adolescent populations. This 'initiation site' is reflected in the set of phenotypes under study that are mainly related to human growth, physical fitness, physical (in-)activity, neuromotor development, bone, and metabolic health.

Main Findings

Findings for distinctive research domains are summarized in Tables 2-5. Studies within the area of growth, physical fitness, physical activity, motor development, bone, and metabolic health require the measurement of young children and adolescents in a school or laboratory setting, with often multiple days of assessment. Therefore, sample sizes are often small in the different studies. Pearson correlation (r), intra-class correlation coefficients (t), or structural equation modeling were used to analyze twin covariation. In most studies, age effects are not modeled as a confounding factor within the structural equation-modeling framework. Given the small sample sizes in most studies, the analytical option was to first regress age and sex from each phenotype, and then compute heritabilities. Had our samples been larger, not only more precise genetic and environmental effects would be computed, but also more firm conclusions would be drawn.

Physical Activity

When using time-consuming interviews, pedometers and accelerometers to assess physical activity measures, our sample sizes are small. In children, genetic factors are small (a^2 from 24% to 34%) for the Godin and Shephard (1985) questionnaire phenotypes (low, moderate, and high intensity, as well as weekly total activity), and in line with the accelerometer data. These results were consistent in mainland and Azorean subjects. In adolescents, questionnaire data (Baecke) showed sex-specific estimates for sport

TABLE 2

Physical Activity

		Saı size	Sample size (pairs)	mple (pairs)			Pearson (r), or intra-class correlation coefficient (t)		Genetic and environmental influences (%)				
Phenotypes	Specific measure	MZ	DZ	Age	Location	MZ	DZ	A	D	С	E	Reference	
Sport participation (Baecke et al., 1982)	Arbitrary unit	40	64	12–18 years	Mainland	Tetrachoric correlations from 0 41–0 95	Tetrachoric correlations from 0 42–0 47	82 (m)			18 (m)	Maia et al. (1999)	
						0.41 0.75	0.42 0.47			71 (f)	29 (f)		
Weekly physical activity interview (Godin & Shephard, 1985)	Arbitrary units	21	52	6–12 years	Azores islands	$t_{MZ} = 0.86$	$t_{DZ} = 0.62$	24			76	Maia et al. (2003)	
School, leisure time, sport participation, and total indices (Baecke et al., 1982)	Arbitrary unit	29	49	11–40 years	Azores islands	t _{MZ} : 0.72–0.92	t _{DZ} : 0.48–0.65	23–77		14–51	8–28	Maia et al. (2001)	
Sport participation, and (SPI) leisure time (LTPA) indices (Baecke et al., 1982)	Arbitrary unit	99	104	11–25 years	Mainland	$r_{MZM} = 0.69$ $r_{MZF} = 0.72$	$r_{\rm DZM} = 0.22$ $r_{\rm DZF} = 0.56$ $r_{\rm DZOS} = 0.31$	63 (m)			37 (m)	Maia et al. (2002)	
	Arbitrary unit					$r_{\rm MZM} = 0.82$ $r_{\rm MZF} = 0.90$	$r_{\rm DZM} = 0.46$ $r_{\rm DZF} = 0.53$ $r_{\rm DZOS} = 0.49$	32 (f) 68 (m)		38 (f) 20 (m)	30 (f) 12 (m)		
5 days monitoring Accelerometer (counts and METs) Pedometer (steps and Kcal)	Counts	32	19	12–18 years	Mainland			40 (f) 34		28 (f)	32 (f) 66	Oliveira & Maia (2002)	
	METs Steps Keal							20	41	90 80	39 10 20		
Weekly physical activity interview (Godin & Shephard, 1985)	LIPA	32	69	6–12 years	Mainland	-	-	34		64	20	Maia et al. (2004)	
(,	MPA VPA							24		82 67	18 9		
5 days monitoring Accelerometer (intensity, from low = till 3 METs, to very vigorous >9 METs)	LPA	18	13	6–12 years	Mainland	r _{MZ} = 0.89	$r_{\rm DZ} = 0.77$					Fernandes & Maia (2006)	
	MPA VPA VVPA					$r_{MZ} = 0.95$ $r_{MZ} = 0.64$ $r_{MZ} = 0.76$	$r_{DZ} = 0.88$ $r_{DZ} = 0.49$ $r_{DZ} = 0.52$						
5 days monitoring Accelerometer (intensity, from low = till 3 METs, to very vigorous, > 9 METs) Pedometer (steps)	LIPA	48	59	10–19 years	Mainland			79 (m)		7 (m)	14 (m)	Sapage & Maia (2007)	
	VVPA							51 (f) 72 (m) 42 (f)		40 (f)	9 (f) 28 (m) 58 (f)		
	Steps							42 (1)		44	12		
5 days monitoring Accelerometer Physical activity patterns: approximate entropy (ApEn)	Counts (ApEn)	77	85	8–18 years	Mainland			44		45	11	Lima et al. (2010)	

Note: m = male; f = female; MZ = monozygotic twin; DZ = dizygotic twin; A = additive genetic effects; D = nonadditive genetic effects; C = shared environmental effects; E = nonshared environmental effects including measurement error.

TABLE 3 Health-Related Physical Fitness

	Specific measure	Sample size (pairs)				Genetic and environmental influences (%)				
Phenotypes		MZ	DZ	Age	Location	A	D	С	Е	Reference
Fitnessgram	1-mile run/walk	21	52	6–10 years	Azores islands	58		24	18	Maia et al. (2001)
		29	42	11–17 years		54		32	14	
	Push-up	21	52	6–10 years		53		31	16	
	•	29	42	11–17 years		80		0	20	
	Trunk-lift	21	52	6–10 years		32		41	27	
		29	42	11–17 years		56		31	13	
	Curl-up	21	52	6–10 years		38		16	45	
		29	42	11–17 years		30		54	16	
Fitnessgram	1-mile run/walk	41	81	6–12 yr	Mainland	50		37	13	Fernandes & Maia (2006)
5	Push-up			,		74		8	18	
	Trunk-lift					34		38	28	
	Curl-up					75		11	14	

Note: MZ = monozygotic twin; DZ = dizygotic twin; A = additive genetic effects; D = nonadditive genetic effects; C = shared environmental effects; E = nonshared environmental effects including measurement error.

TABLE 4

Gross Motor Coordination, and Neuromotor Development

		San Si (pa	nple ze iirs)			Intraclass correlation coefficient (t)		Genetic and environmental influences (%)			
Phenotypes	Specific Measure	MZ	DZ	Age	Location	MZ	DZ	A	С	E	Reference
Gross motor	Backward balance Hoping in one leg Shifting platforms Jumping sideways Global MC	37	27	5–14 years	Mainland	0.73–0.89	0.61–0.69	15–41	46–58	11–32	Chaves et al. (2012)
Neuromotor	Purely motor tasks Dynamic balance Static balance Associated movements	39	56	5–17 years	Mainland	0.42–0.75	0.41–0.56				Lopes et al. (2012)

Note: MZ = monozygotic twin; DZ = dizygotic twin; A = additive genetic effects, D = nonadditive genetic effects, C = shared environmental effects, E = nonshared environmental effects including measurement error.

TABLE 5

Metabolic Syndrome

		Sample size (pairs)				Gen envi influ	etic an ronmer ences (d ntal %)	
Phenotypes	Specific measure	MZ	DZ	Age	Location	А	С	Е	Reference
Metabolic syndrome	WC	84	123	3–18 years	Madeira islands	80		20	Gonçalves et al. (2011)
	SBP					59		41	
	GLU					55		45	
	HDL					34	44	22	
	TRG					61		39	

Note: MZ = monozygotic twin; DZ = dizygotic twin; A = additive genetic effects; D = nonadditive genetic effects; C = shared environmental effects; E = nonshared environmental effects including measurement error.

participation, sports index (males: $a^2 = 63\%$ and $c^2 = 37\%$; females: $a^2 = 32\%$, $c^2 = 38\%$, $e^2 = 30\%$), and leisure time physical activity index ($a^2 = 40\%$, $c^2 = 28\%$, $e^2 = 32\%$), which were coherent with accelerometer low and very vigorous physical activities — in males the genetic additive factors were always greater, and their magnitude was moderate-tohigh (a^2 from 42% to 79\%). When studying physical activity patterns using approximate entropy as a suitable descriptor or a regular pattern of daily activities intensity, genetic factors account for about 50% of the total variation.

Motor Performance

Genetic factors are low to moderate in children's and adolescents' health-related physical fitness (see Table 3).

Endurance running and muscular strength phenotypes had higher a^2 estimates. Gross motor coordination tests presented low genetic influence in children's performance (see Table 4). Twins' MZ and dizygotic (DZ) intra-class correlation coefficients suggest that neuromotor development from childhood to adolescence may be under genetic control, although environmental forces associated with daily living histories and twins' lifestyle may play an important role.

MetS

Within the Madeira Family Study (GEAFAS), DNA was collected for zygosity determination and future association studies with an array of candidate genes. From a vast set of data collected from all three-generation families, MetS information (systolic blood pressure, waist circumference, triglycerides, HDL-cholesterol, and glucose) is available from all subjects. Here, we only report twin data (see Table 5). In all five MetS indicators, MZ had higher intra-class correlations (from 0.56 to 0.81) than DZ twins (from 0.26 to 0.53). The prevalence of MetS (having, at least, any combination of three indicators) was very low: MZ = 2.4%, DZ = 5.3%. A moderate-to-high additive genetic component was found in four of the five MetS indicators (a^2 from 55% to 80%; and e^2 from 20% to 45%), but except HDL-cholesterol ($a^2 = 34\%$, $c^2 = 44\%$, $e^2 = 22\%$).

Future Directions

There is a strong need to merge all of our data sets to address the following issues: (1) to study in more detail physical activity patterns using accelerometer data; (2) to link physical activity levels with health-related physical fitness; (3) to study body composition and its relationship with physical activity levels; (4) to have a careful look at bone phenotypes; (5) to associate MetS indicators with physical activity and physical fitness; (6) to investigate the role of health perceptions in metabolic health and body composition; (7) to study the links of physical self-concept, body composition, and health-related physical fitness. The inter-relations among the phenotypes we examined suggest shared genetic and environmental factors for these phenotypes. In future studies, we plan to apply multivariate twin modeling to the covariations of these phenotypes. The Madeira Islands study includes three-generation families with phenotypes on human physical growth, body composition, biological maturation, physical activity, physical fitness, bone, health-related quality of life, and MetS and has also DNA data collected from 1,200 subjects. As such, an international collaboration is needed to extract the most from this unique data set. Furthermore, a call for education of Portuguese sport and human movement sciences researchers in the field of twin and family genetic methodologies is needed.

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