The Registry of Italian Twin Athletes (RITA): Background, Design, and Procedures, and Twin Data Analysis on Sport Participation—An Application to Twin Swimmers

Paolo Parisi, Benedetta Casini, Valter Di Salvo, Fabio Pigozzi, Monica Pittaluga, Giovanna Prinzi, and Vassilis Klissouras

Aiming to contribute to a better understanding of the nature and characteristics of sport participation and of the factors underlying human performance and adaptation to physical exercise, this study involved (a) the establishment of a specialized twin registry, (b) a questionnaire study, and (c) testing sessions of top level athletes. The general design and procedures of the project are described. A Registry of Italian Twin Athletes (RITA) was developed through a collaborative effort involving 25 sports federations of the Italian National Olympic Committee (CONI). Out of a total of over 1,500,000 registered athletes, some 5,000 pairs of twins practicing sports such as track and field, swimming, tennis, fencing, and cycling, were tentatively identified. A basic questionnaire was devised to address general aspects as well as specific items related to the respondent’s respective discipline, and a specific questionnaire was developed for each sport. As a basis for future studies, an application was conducted on the twins registered with the Swimming Federation. The data collected from the completed questionnaires of about 200 twin swimmers indicated a strong zygosity and sex effect on sport selection, with higher prevalence values for identical pairs and for males. The twins appeared to be taller than their counterparts in the general population, as appears to be true for swimmers in general. Both for anthropometry and basic swimming performance analysis, no zygosity effects were found, individual differences being presumably minimized by basic athletic selection factors. For possible genetic effects to be detected, specific and direct testing sessions on high-level performers should be undertaken.

Key Words: twin registry, sports, genetics, training, physical adaptation, swimming

Key Points:
- Twin research can substantially contribute to clarify the issue of genetic versus environmental determinants of the individual variability in physical exercise, adaptation, and performance, but large and representative samples are needed.
- A population-based registry has been developed, involving over 4000 pairs of twin athletes from 25 sports federations. A questionnaire study has been designed to address general and specific items for each sport, and a first application has been conducted on 200 twin swimmers.
• The data indicate a strong zygosity and sex effect on sport selection, with higher prevalence values for identical pairs and males among the twin swimmers.
• With respect to anthropometry, twin swimmers as a group are taller than their counterparts in the general population, as are swimmers in general. Possible zygosity effects are apparently minimized by the common athletic condition.
• Individual differences in swimming performance and anthropometry being minimized by basic athletic selection factors, possible genetic effects may only be detected by specific and direct testing on high-level performers.

Introduction
Notwithstanding remarkable progress in various areas of human and sports biology, relatively little is known about the respective weight of the genetic and environmental determinants of individual variability in physical exercise, adaptation, and performance. Motor performance is essentially multifactorial in nature, in that its characteristics, and specifically the level it can reach, are the product of a combination of different anatomical, physiological, and psychological traits, that are in turn under the individual and the combined influence of both genetic and environmental factors. Some of these, such as biological features at rest, adaptation to work and to aerobic power, characteristics of muscle and skeletal tissues, muscular strength, motor development and learning, have in the past been the object of genetic investigations conducted with various study models [for reviews see, e.g., Wolanski et al (36), Kovar (18), Klissouras (15)]. Studies of family relatives, and particularly of twins—a basic resource in the attempt to disentangle the variance components involved in the etiology of multifactorial traits [e.g., Parisi (23, 24)]—have also been conducted, along major lines of research that can be broadly subdivided into: (a) general studies on sport participation, motor skills, and learning; (b) biological determinants of motor performance; and (c) response to the training.

Studies on the genetic variation of sport participation, motor development, performance, and learning were already conducted back in the early 1930s [e.g., Holzinger (10), McNemar (21)]. That elite athletes often cluster in families was first stressed some forty years ago by Gedda (3), based on his studies on Italian families of athletes, and confirmed by Grebe with similar studies of German twins (6) and families (7). Following these classic pedigree studies, which indicated the existence of a general family influence, further studies attempted to quantify the role of genetic factors, using the twin concordance model. Gedda’s (4) study of 165 twin pairs took into account aspects such as the kind of sport practiced, specialty or role played, and level of performance. He found concordance for at least two of these in over 80% of MZ, versus only 45% of DZ pairs, and complete discordance in less than 10% of MZ, versus about 30% of DZ pairs. Sample selection factors were not ruled out, however, and may have played a role in contributing to these zygosity differences.

Studies on performance addressed specific fine motor skills, such as finger tapping or other manipulative tasks requiring coordination and dexterity (10, 12, 17, 18, 20, 21, 31), as well as gross motor skills, such as various running, jumping, and throwing tasks (17, 18, 29, 33, 36), and obtained heritability estimates ranging from 0.50 to 0.80 for the former and 0.45 to 0.91 for the latter, with estimates for balance tasks ranging from 0.27 to 0.89 (31, 34, 35). Notwithstanding this large variability in heritability estimates—frequently found in classic twin studies and
stemming from methodological problems, such as the nature of these estimates, ascertainment procedures, sample size, age and sex effects, and so on—there is substantial agreement that MZ twins are significantly more similar than DZ twins in fine motor ability as well as in gross motor development and ability.

Motor learning and exercise must obviously play a role, and presumably influence the above variability, although the data are somewhat conflicting. Marisi (20) found that the heritability for performance on a pursuit-rotor task decreased over 30 practice trials (from an initial 0.79 down to 0.35), in agreement with McNemar’s early findings, back in 1933 (21), that exercise did not modify the degree of similarity of MZ co-twins but increased that of DZ co-twins, thus decreasing heritability estimates. On the other hand, Williams and Gross (34) showed that the heritability for a stabilometer task increased after a two-day practice (from 0.27 to 0.69), and remained close to this final level for the next four days. The genetic component of the learning process should also be considered, and in fact it is likely that motor learning be more similar in MZ than DZ co-twins (30).

As for the biological determinants of motor performance, maximal aerobic power (VO₂max), a global criterion of functional capacity and endurance performance, was found to be under strong genetic control (by Klissouras [13] in 1971), with a heritability estimate of 93% confirmed by comparable estimates obtained in later studies in other laboratories (15). Fairly different results (with heritabilities of about 40% for VO₂max · kg⁻¹ of body mass, and only about 10% for VO₂max · kg⁻¹ of fat-free body mass) were reported by Bouchard et al. (1), while a later path analysis conducted in 1991 by Fagard et al. (2) again estimated heritability at about 70%. A significant genetic variance (with heritabilities of over 70%) has also been estimated for aerobic capacity, determined on the basis of either the total work output during a non-stop 90’ maximal ergocycle test (1) or the lactacid anaerobic threshold at blood lactate concentration of 4 mmol · L⁻¹ (14). Conflicting results were also obtained with respect to the genetic variation of histochemical and biochemical characteristics of the muscle tissue, the heritability of fiber type distribution having been found to be close to unity by Komi and Karlsson (16)—in agreement with Gollnick et al. (5) who had observed no change in muscle fiber distribution following intensive physical training—whereas Bouchard et al. (1) found heritability to be irrelevant. Limited or no genetic effects were also observed for the ultrastructural features of skeletal muscles (mitochondrial volume and density) and for the maximal activity of key regulatory enzymes of glycogen breakdown and substrate oxidation (1, 11, 16).

Finally, with respect to response to the training, this was found to induce adaptation to most biological determinants of sport performance (28), but the finding may be affected by a variety of factors, including the adaptability or “trainability” of the organism. A genotype-training interaction is likely, but few studies have attempted to measure it, and the results are still tentative and somewhat conflicting. In one co-twin control study on mean VO₂max after training (32), 51% of the total intrapair variance was attributed to genotype, 42% to training, and 7% to genotype-training interaction. In a different approach, where both MZ co-twins underwent endurance training (8, 26), resemblance in VO₂max response was found to be high within pairs (0.77) and low among pairs, though largely due to the presence of two extreme cases. Moreover,
improvement appears to be commensurate to pretraining phenotypic levels, which should also be considered (9, 27).

Summing up, the evidence on the role of the various determinants of sport performance is still inconclusive in practically all of the areas considered. This may be due at least partly to methodological problems, such as sample selection and small sample size, as well as to the fact that the twins used in the studies were almost exclusively children or sedentary adults, which means that their subjects had never been challenged maximally, and therefore genetic effects could not be expressed adequately.

It was thought that a better understanding of the role of the variables involved in sport participation and performance might come from studies involving larger samples of twins that are more representative of the general population of twins involved in sport practice. The establishment of a specialized twin registry satisfied these needs and provided a suitable data base for the ascertainment of specific samples of older twins who had already undergone athletic training, including cases of elite twin athletes.

Establishment of the Registry

Primary Ascertainment
A collaborative effort was undertaken involving various research groups of the Rome University Institute of Motor Sciences (IUSM)—previously Higher State Institute of Physical Education (ISEF)—along with 25 Sports Federations of the Italian National Olympic Committee (CONI). The records of the 25 Federations, including a total of about 1,500,000 registered athletes, were obtained and searched. All subjects sharing the same family name and birth date were selected and tentatively assumed to be twins.

The resulting list was computerized and represents the basis of what has been called the Registry of Italian Twin Athletes (RITA). The Registry has initially included a total of 4,628 pairs of assumed twin athletes (2,553 MM, 1,339 FF, 734 MF) practicing sports such as swimming, tennis, fencing, cycling, gymnastics, and others (25).

Secondary Ascertainment
A second ascertainment stage was then planned, involving: (a) a more careful search of the lists in order to reduce overt errors (e.g., the same individual listed twice with slight spelling differences); and (b) crosscheck on local directories, as well as direct contacts with hundreds of individual local clubs, with the aim to confirm the identification of the twins and obtain their addresses. In order to minimize the risk of wrong addresses, the process was scheduled to be completed at the time contact is taken with a specific subsample.

Maintenance
Regular contacts with the federations and local clubs were planned in order to maintain and implement the Registry over the years. This was a very demanding process, and future steps will be largely dependent on funding.
The Questionnaire Study

Development of Questionnaires and Zygosity Determination

A basic questionnaire was devised to address general aspects, such as personal data (date of birth, weight and height, education, etc.), family data (including sports practiced by relatives), and other information. The latter included standard questions meant to allow zygosity determination, such as asking whether the twins, when growing up, were “as alike as two peas in a pod,” whether their mother or other close relative ever mixed them up, and so on. Answers to such questions are held to allow zygosity assessment with a reliability of 95% or over, as estimated by several studies comparing such questionnaire diagnoses to those based on the analysis of blood groups and other genetic markers (23). When direct examination is involved, as in the related studies we are conducting on top-level twin athletes or on elderly twins, DNA fingerprinting is then carried out for maximum reliability. A specific questionnaire section was then devised, in collaboration with experts of the various sports, to address items related to the respective discipline (specialty practiced, training schedule, performance, etc.), diversified for each sport, so that 25 different versions of the questionnaire were developed.

Study Procedures

Each specific questionnaire was planned to be sent with a cover letter to all the twins registered with any given federation, with reminders sent to those who failed to answer after 6 weeks. The attempt was to raise the response rate somewhat. However much improved the quality of the initial data base thanks to secondary ascertainment procedures, the response rate was not expected to be high in light of the outcome of comparable Italian population surveys. The procedures were implemented through a first application to the subpopulation of twin swimmers. Such questionnaire studies were intended to: (a) implement the Registry, and establish a data base of actually available twin pairs in the various sports with a basic assessment of zygosity; (b) collect information, and conduct basic analyses on variables related to personal and family variables of biosocial interest, as well as to anthropometry, lateral dominance, habits and behaviors, and so on; (c) analyze sport participation and identify possible zygosity and sex effects; and (d) identify cases of special interest, and particularly top-level athletes, to be invited to participate in further studies with detailed protocols of direct analyses and testing sessions.

Application: A Study on Twin Swimmers

Materials and Methods

The pilot study on twin swimmers was conceived and designed as a first application of the Registry and the basis for future studies. The questionnaire—with a specific section concerning the style(s) and distance(s) practiced, the records achieved, the training schedule over the last 3 years, possible interruptions of more than 3 months, sport(s) practiced by relatives, and so on—was sent to the 337 twins (167 pairs and 1 set of triplets) identified among the 36,351 athletes registered with the Swimming Federation. The data from the completed questionnaires were descriptively analyzed with the NCSS statistic software package. To allow the comparison between twins, records data were converted into seconds.
Results and Discussion
A total of 194 completed questionnaires were returned, accounting for 88 complete pairs of the various sex combinations (45 MM, 29 FF, and 14 MF), 1 set of MMF triplets, and 15 broken pairs (6 M, 9 F), representing a response rate of ~60%.

Age, Sex, and Family Variables. The subjects were adolescents or young adults, ranging in age from 11.3 to 25.0 (5% trimmed mean = 15.76 ± 3.05). Males considerably exceed females both in the initial population (190 M vs. 147 F) and in the research sample (112 M vs. 82 F). The sex ratio is very similar in the research sample versus the initial group (M:F = 1.36 vs. 1.29), indicating the lack of any considerable sex effect in the participation to the research. No significant differences by sex and zygosity classes were found in birth order (1.5 ± 0.6, range 1–4), family size (4.6 ± 0.8, range 3–7), mean maternal age (28.2 ± 4.2, range 20–41), or paternal age (30.9 ± 4.75, range 19–46) at the birth of twins.

Zygosity and Twinning Rates. Based on the specific questionnaire items, the 88 pairs were classified as follows: 50 monozygotic, or MZ (32 MM, 18 FF), 34 dizygotic, or DZ (10 MM, 10 FF, 14 MF), and 4 of uncertain zygosity, or UZ. As compared to the general twin population rates, there is a much higher frequency of MZ pairs (57% vs. 38%) and same-sex, particularly MM pairs (51% vs. 36%), and a lower frequency of opposite-sex pairs (16% vs. 31%), indicating a zygosity and sex effect on sport selection, and presumably more generally on sport practice. However, the ascertained prevalence of twins among registered swimmers (about 4.6/1000) is much lower than the population twinning rate for the corresponding years (~9.2/1000), indicating that many more individual twins must be present among the 36,351 swimmers registered in the Federation, their co-twins presumably practicing another sport or no sport at all.

Anthropometry. As compared to the general population matched for sex and age class, both male and female individual twin swimmers appear to be significantly taller. In fact, when the individual heights are classified according to Italian standards, over 50% fall between the 25th and the 75th centile, and almost 30% between the 75th and 97th (Table 1).

Table 1   Classification of Twin Swimmers By Mean Height (H) and BMI According to Italian Population Standards: Various Centile Ranges Based on Sex

<table>
<thead>
<tr>
<th>Centile ranges</th>
<th>Total</th>
<th>Male (%)</th>
<th>Female (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>H BMI</td>
<td>&lt;18 yr H BMI</td>
<td>≥18 yr H BMI</td>
</tr>
<tr>
<td>&lt;10°</td>
<td>2</td>
<td>3.9</td>
<td>0</td>
</tr>
<tr>
<td>≥10° to &lt;25°</td>
<td>11.9</td>
<td>15.4</td>
<td>6.5</td>
</tr>
<tr>
<td>≥25° to &lt;75°</td>
<td>59.4</td>
<td>55.1</td>
<td>81.8</td>
</tr>
<tr>
<td>≥75° to ≤90°</td>
<td>11.9</td>
<td>10.2</td>
<td>5.2</td>
</tr>
<tr>
<td>&gt;90°</td>
<td>14.8</td>
<td>16.7</td>
<td>2.6</td>
</tr>
</tbody>
</table>

Although twins are on the average expected to be somewhat smaller than non-twins, this result is not very surprising considering that athletes in general clearly represent a genetically selected and environmentally enriched share of the population in terms of health conditions, fitness, and body
structure. In particular, swimmers as a group are found to be taller and heavier, and with a higher percentage of lean body mass (19, 22).

In the case of body weight (limited to subjects over 18 years old), no significant difference was found in males (72.4 ± 6.3 vs. 71.4), while females appear to be significantly lighter (57.3 ± 5.2 vs. 63.6, \( p < .001 \)). The analysis of the body mass index (BMI) indicated 84% of the males and 79.7% of the females fall between the 25th and 75th centile. Females appeared to be more underweight, with 20.3% under the 25th centile, and 16.3% of those younger than 18 years under the 10th centile (Table 1), which may reflect a tendency common among girls of that age, particularly athletes.

Considering the whole sample, intrapair correlations are similarly high in MZ and same-sex DZ co-twins, both for height (0.97 vs. 0.95) and weight (0.95 vs. 0.91) as well as for BMI (0.86 vs. 0.79) (Table 2). This must also be due to the above-mentioned athletic selection effect, minimizing individual differences and therefore increasing intrapair correlations.

### Table 2  Intrapair Correlations for Height, Weight, and BMI in Swimmers By Zygosity

<table>
<thead>
<tr>
<th></th>
<th>Height</th>
<th></th>
<th>Weight</th>
<th></th>
<th>BMI</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>MZ</td>
<td>DZ same-sex</td>
<td>MZ</td>
<td>DZ same-sex</td>
<td>MZ</td>
</tr>
<tr>
<td>Pairs (n)</td>
<td>47</td>
<td>20</td>
<td>45</td>
<td>19</td>
<td>45</td>
</tr>
<tr>
<td>Mean of twin A</td>
<td>166.8 ± 9.9</td>
<td>161.0 ± 15.1</td>
<td>56.2 ± 11.2</td>
<td>51.7 ± 16.4</td>
<td>19.9 ± 2.2</td>
</tr>
<tr>
<td>Mean of twin B</td>
<td>166.8 ± 10.1</td>
<td>161.9 ± 13.2</td>
<td>56.9 ± 11.8</td>
<td>53.0 ± 15.0</td>
<td>20.2 ± 2.4</td>
</tr>
<tr>
<td>r</td>
<td>0.97</td>
<td>0.95</td>
<td>0.95</td>
<td>0.91</td>
<td>0.86</td>
</tr>
</tbody>
</table>

### Table 3  Intrapair Correlations for Training Schedule in the Twin Swimmers By Zygosity

<table>
<thead>
<tr>
<th></th>
<th>Years of training</th>
<th>Months/year</th>
<th>Sessions/week</th>
<th>Hours/session</th>
</tr>
</thead>
<tbody>
<tr>
<td>MZ</td>
<td>Pairs (n)</td>
<td>43</td>
<td>34</td>
<td>33</td>
</tr>
<tr>
<td></td>
<td>Mean of twin A</td>
<td>6.1 ± 3.8</td>
<td>9.7 ± 1.0</td>
<td>5.3 ± 1.1</td>
</tr>
<tr>
<td></td>
<td>Mean of twin B</td>
<td>5.9 ± 3.8</td>
<td>9.7 ± 1.0</td>
<td>5.2 ± 1.1</td>
</tr>
<tr>
<td></td>
<td>r</td>
<td>0.96</td>
<td>0.74</td>
<td>0.95</td>
</tr>
<tr>
<td>DZ</td>
<td>Pairs (n)</td>
<td>28</td>
<td>12</td>
<td>12</td>
</tr>
<tr>
<td></td>
<td>Mean of twin A</td>
<td>5.4 ± 3.6</td>
<td>10.0 ± 0.8</td>
<td>5.1 ± 0.8</td>
</tr>
<tr>
<td></td>
<td>Mean of twin B</td>
<td>5.2 ± 3.2</td>
<td>10.1 ± 0.7</td>
<td>5.2 ± 0.9</td>
</tr>
<tr>
<td></td>
<td>r</td>
<td>0.92</td>
<td>0.71</td>
<td>0.97</td>
</tr>
</tbody>
</table>

**Sport Performance Analysis.** Performance was studied under various aspects, from the stroke chosen to perform in competitions, considering first and second choice, to the rating of interruption of competition activity, other sports practiced, training pattern, and personal record. With respect to stroke, intrapair correlation rates are lower than expected in MZ pairs, and only slightly higher than in DZ pairs for the first choice (38% vs. 26%) but not the second (25% vs. 26%). This is probably owing to the fact that the stroke choice at a competitive level is generally made by the coach on the basis of the best yield. High concordance values are instead found in
both MZ and DZ pairs for interruption rates (72% vs. 80%), and partly for the choice of a secondary sport (66.7% vs. 50%).

The training patterns (considering months per year, sessions per week, and hours per session) are highly correlated in both zygosity groups (Table 3). This may also reflect the fact that in competitive practice, the training schedule basically is fixed by the coach. The high correlation for the number of years of training is obviously a consequence of the fact that the twins are usually introduced in sport practice at the same time.

With respect to performance, we could only compare those pairs in which both twins race the same stroke on the same distance, and most of these participate in the 100-m freestyle, probably the least influenced by the learning process. Intrapair correlations between records are almost close to unity in both zygosity groups. Splitting all pairs and rearranging the subjects at random, while maintaining the correspondence for stroke, distance, and sex, the correlation is lower (0.75) (Table 4).

<table>
<thead>
<tr>
<th>Table 4</th>
<th>Intrapair Correlations for Performance Between Same-Sex Twins Practicing the Same Stroke on the Same Distance</th>
</tr>
</thead>
<tbody>
<tr>
<td>Variable</td>
<td>Personal record (sec)</td>
</tr>
<tr>
<td>Pairs (n)</td>
<td>14</td>
</tr>
<tr>
<td>Mean of twin A</td>
<td>68.7 ± 30.0</td>
</tr>
<tr>
<td>Mean of twin B</td>
<td>69.1 ± 29.3</td>
</tr>
<tr>
<td>r</td>
<td>0.99</td>
</tr>
</tbody>
</table>

Discussion

The establishment of a twin registry specifically devoted to twins involved in sport practice represents a research tool that may prove of great use for a variety of studies in human genetics and sport science. This is particularly true in view of the number of participants involved, which allowed the study to overcome the usual problem of limited twin sample size. Of great methodological importance is also the lack, or limited effect, of ascertainment bias—the registry being a reference to what should be considered a statistical population rather than a sample, with no specific selection criteria other than sport practice and registration with a national sports federation. The result is the availability of considerable numbers of nonselected twin pairs practicing the various sports, thereby avoiding many of the methodological problems usually encountered in twin research (23, 24).

The basic data collected for the establishment of the registry and the specific application to the subpopulation of twin swimmers already lend themselves to some considerations and indications that may prove useful for future work.

A first point concerns sport participation. It has long been held (4, 7) that sport participation, as well as concordance for specialty selection and levels of performance, are much higher in MZ than DZ pairs. This finding is only partly confirmed by present data, which rather seem to indicate that nonrandom sampling, based on the study of specific twin pairs, may have led to
overestimating the prevalence of MZ pairs and their intrapair concordance in previous sample studies.

In fact, the number of twin pairs found in the total population of registered athletes (4,626 in 1,500,000 or 3.1/1000) represents about one third of what should be expected based on the prevalence of twins in the general population (about 9.2/1000). In the subpopulation of swimmers that was more directly studied, the ratio was slightly higher (167 in 36,351, or 4.6/1000) but still less than half of what should be expected. It must be concluded that many broken pairs exist, with co-twins practicing a different sport or no sport at all.

Twin pairs (but not necessarily twins) are therefore underrepresented among athletes, which is after all not surprising. However, within the twin pairs, among swimmers, the proportion of MZ pairs is higher than expected (57% vs. 38% in the general twin population), suggesting that some zygosity effect does exist in sport participation. Finally, and as expected, males are also overrepresented among swimmers.

As for intrapair comparisons, and contrary to what one might expect, these generally fail to indicate higher similarity values in MZ than DZ pairs. This is true for anthropometry as well as for sport performance and training patterns among our twin swimmers. Intrapair correlations are indeed high, but more or less equally so in MZ and DZ pairs, thus failing to support relevant genetic effects. This may be due to selection factors, such as the fact that the subjects had all chosen swimming and practiced it for years, minimizing interindividual differences, at least when such general variables are concerned. In fact, mean values and standard deviations are quite similar in the various groups.

Even so, when individual twin subjects are combined in random pairs of nonrelated individuals and compared with respect to performance, the correlations found in such random pairs are considerably lower ($r = 0.75$ vs. $r = 0.99$ in twin pairs), indicating that family factors do play a role. However, for specific genetic effects to be detected, the analysis should necessarily focus on traits characterized by wider variability.

References

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Vassilis Klissouras is an exercise physiologist, with particular interest in twin research as applied to the genetic basis of adaptive variation and to the biological determinants of human performance. Professor at McGill University, Montreal, Canada (1967–1984) and, since then, at the University of Athens, where he has also been the founding Dean of the Faculty of Physical Education and Sport Science. Fellow of the American College of Sport Medicine and Founding Fellow of the European College of Sport Science.