Contribution of genetic and environmental effects to postural balance in older female twins

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GOOD POSTURAL BALANCE IS REQUIRED for carrying out everyday tasks. With increasing age, sensory and motor control of posture is often affected (32, 57). This can potentially lead to perturbations when one is standing and even to injurious falls. Knowledge about factors behind individual differences of postural control during aging is limited. Furthermore, it is not known how big a proportion of the variability in postural balance in older persons.

METHODS

Participants. The present study is a part of the Finnish Twin Study on Aging (FITSA), a study on contribution of the genetic and environmental influences on the disablement process in older women. FITSA participants were recruited from the Finnish Twin Cohort (24, 25), which comprises all same-sex twin pairs born before 1958 and alive in 1975. In the age group of 63–76 yr, there were 1,260 respondent female pairs. Among them, those who had participated in Finnish Twin Cohort Study in 1975 were included when the sample for the present study was selected. To obtain study groups with equal number of monozygotic (MZ) and dizygotic (DZ) twin pairs, an invitation to participate in the study was sent, in August 2000, to 178 MZ pairs, 212 DZ pairs, and 24 pairs of previously undetermined zygosity (XZ). Inclusion criteria were the willingness of both twin sisters to participate and the self-reported ability to walk 2 km and to travel independently to the laboratory. Reasons for nonparticipation were refusal (50 MZ, 51 DZ, and 5 XZ pairs), poor health status (28 MZ, 52 DZ, and 5 XZ pairs), or death (2 MZ, 3 DZ, and 1 XZ pairs) of one or both twin sisters. Finally, 98 MZ, 106 DZ, and 13 XZ pairs arrived at the laboratory. The twin pairs had been initially determined as MZ, DZ, or XZ twins in 1975 by using a questionnaire in which both twins answered questions about their similarity of appearance in childhood. This method correctly classifies 93% of twins with a 1.7% probability of misclassification (42). During the present study, the zygosity of the 13 XZ pairs was ascertained by 10 highly polymorphic genetic markers at the National Public Health Institute Paternity Laboratory. Four pairs

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were determined as MZ and nine pairs as DZ twins. Thus the final sample consisted of 102 MZ pairs and 115 DZ pairs; the zygosity of the DZ pairs was correctly determined as part of an ongoing genome scan. On arrival at the laboratory, the participants gave their written, informed consent.

Postural balance measurements. Postural sway tests were done as a part of a battery of laboratory examinations that lasted ~5 h per participant and included measurements of muscle strength, bone mineral density, eye sight, hearing, pulmonary capacity, and tests of functional ability. Sway tests were done for all participants in the afternoon, after a 30-min rest interval, and were carried out by two physiotherapists.

Sway was measured with the subject standing on stocking feet in a given position on the force platform (Good Balance, Metitur). During the tests, the participants were instructed to stand as still as possible in a well-balanced position. For the side-by-side stance tests, participants were requested to self-select a natural and comfortable standing position on the force platform. Typically, a participant stood with her feet 15–25 cm apart. The test was done first with the eyes open (side-by-side EO) and then repeated with the eyes closed (side-by-side EC). The semi-tandem test was performed with the eyes open (semi-tandem EO) and with one foot placed one-half a foot length ahead of the other, with feet touching. Participants were allowed to place their hands on the triangular-shaped platform because the midpoint for each test was calculated on the basis of the data obtained from that particular measurement. In both side-by-side tests, the arms were held in a relaxed position in front of the body with the hands clasped together. During the semi-tandem EO test, participants were asked to keep their arms down by their sides. Gaze was fixed at a marked point at eye level at the distance of 2 m. In the side-by-side EO and EC tests, sway was recorded for 30 s and in the semi-tandem EO test for 20 s. Recording started when a balanced and safe stance had been attained. One trial was performed in each test, and between tests participants were allowed to sit down and rest for a few minutes. Tests were done in the same order, from easiest to more difficult, for all participants.

The Good Balance system comprises a portable triangular (800 × 800 × 800 mm) force platform with strain-gauge transducers connected to a three-channel direct-current amplifier and 12-byte analog-to-digital converter connected to a computer. In the present study, the sampling frequency for each channel was 50 Hz. After all measurement points were read, the medio-lateral (x) and anteroposterior (y) coordinates of the center of pressure (COP) were calculated on the basis of these vertical force signals by using the Good Balance software. The error in the calculated x and y coordinates of the COP is <1.0 mm when the mass of the subject is at least 40 kg (34a). The device was calibrated weekly.

Three outcome variables for each test were chosen for the further analyses. On the basis of the displacement of the COP, the length of the path that the COP traveled during each second of the test in the mediolateral (ML) and anteroposterior (AP) directions was indicated by two variables: ML velocity (mm/s) and AP velocity (mm/s) (17). The mean moment of velocity (VEL; mm²/s) was calculated as the mean of the areas covered by the movement of COP during each second of the test (17). To compensate for the possible influence of higher locations of the center of mass among the taller subjects, absolute sway measures were adjusted for the subject’s height [(sway variable/subject height) × 180] (17).

Background information on health and physical activity. Participants’ body height and weight were measured by a nurse in the laboratory. The questionnaire used to assess present status of physical health, diseases, smoking, and difficulties in walking was based on the questionnaires used in the earlier epidemiological (18) and twin studies (25). Participants indicated the chronic conditions they had from the following list: coronary heart disease, cardiac failure, hyper-tension, pulmonary diseases, asthma, multiple sclerosis, Parkinson’s disease, arthrosis, rheumatoid arthritis, fibromyalgia, gout, deficiency or hyperactivity of the thyroid gland, diabetes, and cancer. Disease status was ascertained during a clinical examination by a physician. Data on the use of prescribed medication was obtained by interviewing the subjects and noting prescriptions. With the use of the scale of physical activity level developed by Grimby (17a), participants were classified as sedentary, indicating that no other regular activity but light walking two or fewer times a week was performed, moderately active, meaning walking or other light exercise was performed at least three times a week but no exercise more intensive than that, or active, when moderate or vigorous exercise was engaged in at least three times per week.

Statistical analysis. Complete postural sway data were obtained from 97 MZ and 112 DZ pairs. Missing values were replaced for those individuals who lacked the result for a single test, providing her sister had complete data for all three tests (5 MZ and 4 DZ individuals). The main reason for not obtaining the test result was that the test was not completed because of the pain in the lower extremity or a lack of concentration on the test leading to loss of balance. Missing values were inputted with the values calculated by using the expectation maximum algorithm method. This method takes into consideration the structure of entire data when the values are estimated (34). The final sample consisted of 101 MZ and 114 DZ pairs. The equality of the mean sway results between MZ and DZ twin individuals was tested with the multivariate analysis of variance (MANOVA). The Kolmogorov-Smirnoff test was used to assess the normality of the data. After natural logarithmic transformation, all the sway parameters showed normal distributions. Intraclass correlation coefficients were computed for the MZ and DZ pairs with the SPSS (45). Comparison of the correlations between the MZ and DZ pairs provided indicative estimates of the importance of genetic and environmental influences on postural sway.

Because the postural sway variables correlated considerably across test situations (Table 1) and the preliminary factor analysis showed that these variables loaded onto three factors, a measurement model was constructed to condense the information. First, an 18 × 18 matrix, containing variances and covariances of nine sway variables for both twin sisters and covariances of the sway variables between the sisters, was calculated by using the PRELIS 2.52 (22). A measurement model was then constructed by using the multisample method of the LISREL 8.5 (23). In the model, factor loadings, variances of the factors, and residual variances were set equal for both twin sisters across the MZ and DZ groups. This was done to keep the structure of the measurement model similar in the MZ and DZ groups. Finally, the constructed model was fitted to the observed covariance matrix by using the maximum likelihood method, and the fit of the model was evaluated by using the χ² test (28). In addition to the χ² test, use of several fit indexes is recommended to assess the fit of the model to the data (7, 28). In the present study, root mean square error of approximation (RMSEA) (47), comparative fit index (CFI) (19), and nonnormed fit index (NNFI) (5) were used, and the rules suggested by Hu and Bentler (20) (RMSEA < 0.06, CFI > 0.95, and NNFI > 0.95) were applied.

To determine the relative contribution of genetic and environmental influences to the estimated variance of the latent factors in the measurement model, structural equation modeling was used. Genetic modeling is based on the fact that MZ pairs share all of their genes in common by descent and that DZ pairs share on average 50% of their segregating genes. A further assumption is that MZ and DZ twins are equally susceptible to environmental influences that are productive of similarities between both twins. Therefore, observed phenotypic variance can be decomposed into additive (A) and dominant (D) genetic influences, the effects of environmental influences that are shared by each twin (C), or individual factors unique to each twin (E). In the present data, the pattern of MZ and DZ twin pair correlations suggested no effect of D, which was therefore not included in the models. The ACE model was fitted to the latent factors of the measurement model. Then, A, C, or E was dropped one at a time, and the overall fit
of the model with the data was tested by the χ² test. Comparison of the submodels was done by using the χ² difference test (5) and Akaike’s information criterion (AIC) values (1). Our goal was to find the most parsimonious model that statistically fitted the data well while also providing a theoretically meaningful interpretation.

**RESULTS**

No significant differences were observed between MZ and DZ twin individuals in age, body height, body mass, self-rated health and physical activity, difficulty in walking, smoking, number of prescription medicines, or sway variables. The number of chronic conditions and prevalence of specific diseases did not differ between MZ and DZ twins (Tables 2 and 3). Table 3 also shows how the increase in the difficulty of the stance was reflected in increased values in the sway variables measured. For example, the lowest velocity moment was recorded for the side-by-side stance with eyes open and the highest for the semitandem test. Table 4 shows that most of the within-pair correlations were higher in the MZ than DZ pairs, suggesting that additive genetic factors (A) contributed to variability. For a few sway variables, however, the correlations were relatively strong but almost equal, indicating the importance of a shared environment (C), which enhances similarity in twins, irrespective of zygosity.

**Measurement model.** The construction of the measurement model started with the formation of three latent factors named side-by-side EO, side-by-side EC, and semitandem EO. Each factor incorporated all three sway variables measured in that particular stance. Because the side-by-side EO, side-by-side EC, and semitandem EO factors correlated considerably with each other, a second-order factor, named balance, was added to the model. Furthermore, the residual correlations found between the AP velocity variables indicated the need for an additional specific latent factor, named AP sway. Each latent factor for twin A and comparable factor for twin B, in the MZ and DZ groups separately, were allowed to correlate. In this model (Fig. 1), within-pair correlations for first-order factors side-by-side EO and side-by-side EC were observed not to be statistically significant in either the MZ or DZ groups and,

<table>
<thead>
<tr>
<th>Variable</th>
<th>MZ Twin Individuals (n = 202)</th>
<th>DZ Twin Individuals (n = 228)</th>
<th>P Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age, yr</td>
<td>68.2 ± 3.7</td>
<td>69.9 ± 3.1</td>
<td>0.14</td>
</tr>
<tr>
<td>Body height, cm</td>
<td>158.3 ± 6.3</td>
<td>159.0 ± 5.9</td>
<td>0.34</td>
</tr>
<tr>
<td>Body mass, kg</td>
<td>69.6 ± 11.6</td>
<td>70.6 ± 12.3</td>
<td>0.51</td>
</tr>
<tr>
<td>Prescription medication, n</td>
<td>1.9 ± 2.1</td>
<td>2.1 ± 1.9</td>
<td>0.23</td>
</tr>
<tr>
<td>Chronic conditions, n</td>
<td>2.0 ± 1.5</td>
<td>1.9 ± 1.4</td>
<td>0.56</td>
</tr>
<tr>
<td>Self-rated health, %</td>
<td>0.28</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Poor/fair</td>
<td>6</td>
<td>4</td>
<td>0.14</td>
</tr>
<tr>
<td>Good</td>
<td>63</td>
<td>71</td>
<td>0.34</td>
</tr>
<tr>
<td>Very good/excellent</td>
<td>31</td>
<td>25</td>
<td>0.51</td>
</tr>
<tr>
<td>Physical activity, %</td>
<td>0.66</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Sedentary</td>
<td>27</td>
<td>28</td>
<td></td>
</tr>
<tr>
<td>Moderate active</td>
<td>50</td>
<td>53</td>
<td></td>
</tr>
<tr>
<td>Active</td>
<td>23</td>
<td>19</td>
<td></td>
</tr>
<tr>
<td>Difficulty in walking 2 km, %</td>
<td>0.73</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Presently smoking, %</td>
<td>0.10</td>
<td></td>
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Values are means ± SD of age, body mass and height, and number (n) of self-reported chronic conditions and prescription medications. Self-rated health, physical activity level, difficulty in walking 2 km, and present smoking are given as percentages. P values indicate no significant (P < 0.05) differences between MZ and DZ groups.
consequently, these correlations were set at zero. The model obtained was accepted as the final model because the fit indexes CFI = 0.96, NNFI = 0.96, and RMSEA = 0.06 showed good fit, although the \( \chi^2 \) test was significant \( [\chi^2(310) = 432.50, P = 0.0001] \). The completely standardized model with parameter estimates and standard errors is presented in Fig. 1. Because age showed no significant association with the latent factors in the model, it was not included in further analyses.

**Genetic and environmental variance.** Genetic influences were suggested by the higher within-pair correlations in the MZ compared with DZ pairs for the balance and AP sway factors (Table 5). The results of fitting A, C, and E to the measurement model factors are shown in Table 6. Path coefficients and fit indexes for the ACE models and reduced models indicated only minor differences in the fit of the models. The model best describing the present data is presented in Fig. 2. In this model, A accounted for 35% and C accounted for 24% of the total variance in the balance factor. Variation of AP sway factor, however, was accounted for by genetic (51%) and individual environmental influences (49%). For the semitandem EO factor, genetic and environmental influences contributing to the variance were partially provided through the balance factor. Moreover, genetic, shared and individual environmental effects specific to the semitandem EO factor were found; these accounted for 11, 16, and 38% of the variance, respectively.

**DISCUSSION**

In the present study, genetic effects accounted for one-third and shared environmental influences for one-fourth of the total variance in the balance factor, which describes the phenotype of postural balance by condensing data from multiple postural sway measurements. To the best of our knowledge, this is the first study on genetic and environmental influences on postural sway among older women.

In an earlier study, Carmelli et al. (10) found no evidence of twin-pair similarity in the ability to hold three progressively more difficult stances for a given time (side-by-side stance, semitandem stance in which the heel of one foot is placed...
beside the first toe of the other foot, and a tandem stance in which one foot was completely in front of another). Performance was evaluated by using a composite score ranging from 0 (not able to stand in any of the positions) to 4 (able to hold all required positions). Mean score was 3.77 (standard deviation of 0.58) for MZ twins and 3.70 (standard deviation of 0.69) for DZ twins, suggesting that almost all participants got the maximum score. Consequently, the lack of variability in the test score may have impaired the estimation of variance components in that study.

In our study, the ACE model was selected because it provided a physiologically meaningful explanation for the sources of variance in the balance and semitandem EO factors. However, the statistical power of the present study, owing to sample size, did not allow us to indisputably confirm the superiority of the ACE model over the other models. The main reason for choosing the ACE models was that the bodily structures and functions involved in postural control, such as the central nervous system, information processing speed, tactile sensitivity, and body size are known to have high to moderate genetic influences, suggesting that there are multiple plausible genetic mechanisms contributing to postural balance.

With increasing age, the capacity of the central nervous system to appropriately process the afferent signals from multiple sources may become a limiting factor in the control of an upright posture (33, 49). Therefore, the genetic influences on the central nervous system may at least partially underlie the genetic influences on postural balance. For psychomotor speed, indicated by reaction time in simple and multiple-choice tests, the present data suggested the contribution of genetic influences, although moderately (16). However, for information processing speed, indicated by the digit symbol test, previous studies have suggested the heritabilities between 68 and 85% in older people (41, 48). In the central nervous system, the cerebellum plays a central role in controlling standing posture. In relatively unchallenging conditions, such as during quiet stance on a stationary platform, peripheral sensation has a significant role in maintaining balance (9, 17, 29). In the present study, the balance factor captured the variance of the first-order factors, regardless of whether the sway was measured with the subject keeping the eyes open or closed. It is likely, therefore, that this factor represents the aspect of pos-

<p>| Table 6. Results of fitting the genetic model for latent factors balance, semi-tandem eyes open, and anteroposterior sway, fit index and path coefficients |
|---------------------------------|------------------|------------------|------------------|------------------|------------------|</p>
<table>
<thead>
<tr>
<th>Factor and Fitted Model</th>
<th>df</th>
<th>Fit indexes</th>
<th>AIC</th>
<th>C</th>
<th>E</th>
</tr>
</thead>
<tbody>
<tr>
<td>Semi-tandem</td>
<td>310</td>
<td>187.46</td>
<td>0.59</td>
<td>0.49</td>
<td>0.49</td>
</tr>
<tr>
<td>Semi-tandem</td>
<td>311</td>
<td>189.46</td>
<td>0.59</td>
<td>0.49</td>
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</tr>
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<td>Semi-tandem</td>
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<td>190.27</td>
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<td>0.59</td>
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tural balance that is maintained by using control systems other than vision, such as tactile sensitivity.

Distal sensory loss that may have a debilitating effect on postural balance control is usually caused by peripheral neuropathies (6). Some of these neuropathies are monogenetically heritable, and mutations in 17 genes have been identified as underlying these diseases (27). More commonly, however, peripheral neuropathies are secondary to diabetes (50). For example, TNFRSF1B (3) and the presence of the ATP1A1 gene variant (55) have been identified as candidate genes that predispose their carriers to diabetic neuropathy. However, among nonclinical populations, only limited information about the heritability of tactile sensitivity exists. Among the participants of the present study, the higher MZ than DZ correlation of vibrotactile threshold in the foot (results not shown) suggests that genetic influences account for at least some of the variability in sensation perception among the population at large. It is plausible that part of the genetic mechanism governing postural balance may be mediated through genetic influences on tactile sensitivity.

Muscles play a central role in maintaining an upright stance. The coordination and timing of corrective movements are essential, but muscle force is produced at submaximal levels. There are very few studies on the heritability of lower extremity muscle strength characteristics in older people. In the present study sample, 33% (95% CI = 19–45%) of the individual differences in maximal isometric knee extension strength were accounted for by additive genetic effects (53). Arden and Spector (2) reported that 46% (95% CI = 3–69%) of the variance in maximal leg extension power was accounted for by genetic influences. It is probable that, as the stance position increases in difficulty and in the presence of impairments in other postural control systems, the relative importance of muscle force in controlling an upright posture also increases. Even though at present no evidence exists to show that muscle strength and postural stability share a genetic component, this mechanism is a possibility and warrants further study.

Body height and mass have been demonstrated to be under strong genetic control (26, 43). Greater body size increases the challenge for postural control systems (17). In the present study, even after adjusting the sway results for height, the sway variables of the more difficult standing positions correlated with height, whereas the anteroposterior sway variables correlated with body mass. Thus genetic effects on body size may be one of the sources of genetic influences explaining variability in postural balance.

In our analysis, we condensed data obtained from multiple sway measurements to fewer factors. Our model demonstrated that the majority of the variance of the sway variables was explained by three first-order factors that loaded onto one second-order factor. However, a variance that was specific for stance position or sway direction and accounted for separate
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Genetic and environmental influences was also observed. This finding suggests that situation-specific mechanisms probably have their own underlying genetic mechanisms. To comprehensively measure a complex trait such as postural balance, therefore, it is essential to use multiple tests in different standing positions.

To be recruited to the present study, both sisters had to agree to participate and be able to travel independently to the research laboratory. Therefore, the present sample was composed of relatively healthy and mobile older women. However, persons with poor mobility and possible related balance impairments were more likely to drop out, which, at least to some extent, reduced the variance in sway values.

Our study was the first to estimate genetic and environmental influences on postural balance in a group of older women. The present study showed that individual environmental factors accounted for up to one-half of the variability of the sway indicators. Because heritability is a population- and gender-specific estimate (39), the present results cannot be directly generalized to other population subgroups. There is a clear need for further studies to elucidate the separate role of genetic and environmental influences on balancing ability in more challenging situations and in frail populations. Even though some people may generally be more disposed to have poor postural balance, placing them at increased risk for falls and mobility limitation, modification of environmental factors, such as physical activity, has great potential. It is worth noting that, in particular, those with poorer “starting values” may benefit most from interventions.

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REFERENCES


