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Genetic and environmental influences underlying the relationship between autistic traits and temperament and character dimensions in adulthood

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Abstract

Background: In recent years, several twin studies adopted a dimensional approach to Autism Spectrum Disorders (ASD) and estimated the contribution of genetic and environmental influences to variation in autistic traits. However, no study was performed on adults over 18 years of age and all but two studies were based on parent or teacher ratings. Also, the genetic and environmental contributions to the interplay between autistic traits and adult personality dimensions have not been investigated.

Methods: A sample of 266 complete twin pairs (30% males, mean age 40 ± 12 years) drawn from the population-based Italian Twin Register was administered the Autism-Spectrum Quotient, Temperament and Character Inventory (TCI-125), and General Health Questionnaire (GHQ-12). Genetic structural equation modelling was performed with the Mx program. Estimates were adjusted for gender, age, and GHQ-12 score.

Results: Genetic factors accounted for 44% and 20%–49% of individual differences in autistic traits and TCI dimensions, respectively. Unshared environmental factors explained the remaining proportion of variance. Consistently with the notion of a personality profile in ASD characterised by obsessive temperament, autistic traits showed significant phenotypic correlations with several TCI dimensions (positive: HA; negative: NS, RD, SD, C). Genetic and unshared environmental correlations between AQ and these TCI dimensions were significant. The degree of genetic overlap was generally greater than the degree of environmental overlap.

Conclusions: Despite some limitations, this study suggests that genetic factors contribute substantially to individual differences in autistic traits in adults, with unshared environmental influences also playing an important role. It also suggests that autistic traits and the majority of temperament and character dimensions share common genetic and environmental aetiological factors.

1. Introduction

Autism Spectrum Disorders (ASD) are a set of phenotypically heterogeneous neurodevelopmental syndromes characterised by difficulties in social communication and social interaction, and unusually restricted, repetitive behaviours and interests [1]. These disorders are considered to be highly heritable. In twin studies, the concordance in monozygotic twins is much higher than that in dizygotic twins [2]; also, the individual risk of ASD was found to increase with increasing genetic relatedness [3].

Rather than treating ASD as discrete entities with a categorical approach based on a distinct boundary between normality and pathology, several authors [4,5] have suggested a dimensional approach to ASD, which conceptualises these disorders as the upper extreme of a constellation of deficits in social adaptation and communication that may be continuously distributed in the population. There are several lines of evidence for this notion: autistic traits measured in the general population show a smooth distribution throughout the normal range to the clinical extreme [6,7]; relatives of patients with ASD display high levels of autistic traits [8,9]; factor analytic approaches

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did not detect discontinuities between ASD and autistic traits [10]; known risk factors for ASD (e.g., paternal age at birth) have been shown to influence autistic traits [11]; common genetic variants that are, by their very nature, present in a significant proportion of the general population, are believed to play a role in the aetiology of ASD [12–14]. Indeed, the DSM-5 itself has highlighted the dimensional nature of the cardinal behavioural domains of ASD, by incorporating a severity scale to capture the 'spectrum' nature of ASD [1].

The dimensional approach to ASD has several potential advantages, such as enhancing the statistical power of genetic studies and averting the problem of misclassification, and it has also been incorporated in twin studies. The large majority of these studies were performed on children and young adolescents, and were based on parent or teacher ratings; this is an important methodological aspect because external and self-rated reports may yield diverse results, as different raters can provide dissimilar perspectives on behaviour [15,16]. The studies performed in early childhood yielded moderate heritability estimates (40% and 44%) for parent-rated autistic traits [17,18]. In older children and young adolescents, most studies reported high heritability (60%–90%) for parent- and teacher-rated autistic traits [6,19–25], while heritability was found to be moderate (36%–47%) for self-reported autistic traits in 9-year-old children [23]. These studies consistently reported moderate influences of the unshared environment, whereas they did not yield a consistent picture regarding the influence of the shared environment; most studies found no significant effects, while some studies in early [17,18] and middle-to-late childhood [20,23,24,26] reported modest (4%–32%) shared environmental influences. Of importance, genetic and environmental influences were found to be stable in the population with increasing levels of autistic traits [24]; also, a strong correlation, predominantly affected by genetic factors, was observed between narrowly defined extreme-end conditions and the full variation of autistic traits [25]. A study on late adolescents aged 18 years corroborated the pattern of results observed at earlier ages, as it reported substantial heritability (57%) and moderate unshared environmental influences (43%) on self-reported autistic traits [27].

To our knowledge, only two twin studies [23,27] were based on self-report ratings of autistic traits, and no study was performed on adults over 18 years of age. Such a study would have the potential to increase the understanding of developmental change and continuity in genetic and environmental influences on ASD and autistic traits. Also, the genetic and environmental influences on the interplay between autistic traits and higher-order adult personality dimensions have not been explored. Moreover, no twin study of autistic traits took account of the possible influence of state variables such as emotional distress, which has been found to be correlated with scores on autistic trait measures [28,29].

The present study aimed at filling these research gaps by providing a reliable estimate of the genetic and environmental components of autistic traits in adulthood, and by exploring if and to what extent these components are related to those of higher-order personality dimensions. We used a multivariate, genetically informed twin design in a general population sample in order to (1) explore the genetic and environmental architecture of autistic traits in adults while adjusting for emotional distress; (2) investigate the relationship between autistic traits and higher-order personality traits; (3) unravel the genetic and environmental bases of this relationship.

2. Methods

2.1. Subjects

The study subjects were drawn from the population-based Italian Twin Register (ITR). The procedures that led to the establishment of the ITR are described in detail elsewhere [30]. Currently, the ITR includes more than 25,000 twins, and is involved in both general population and clinical studies on a large variety of complex phenotypes, with behavioural and psychiatric genetics as major research areas [31].

This study is embedded within a broad mail survey on health and psychological well-being in adulthood, targeting three metropolitan areas of Northern (Milan), Central (Rome) and Southern (Palermo) Italy. From February to November 2010, twins aged 18–65 years, previously enrolled in the ITR, were invited to participate in the survey and were asked to donate a saliva sample to be stored for future genetic and epigenetic studies. In the same contact, the questionnaires on the traits of interest were sent to the twins. After excluding unmatched twins, a total of 532 subjects from 266 complete twin pairs were left for the analysis. Their mean age was 39.9 years (SD = 11.9; range 18–65). About one third (N = 158, 30%) were males, while two thirds (N = 374, 70%) were females. Most participants had high school (54%) or university (34%) education. About three quarters of them (76%) worked part- or full-time, while 7% were housewives, 4% were still studying, 3% were unemployed, and 10% did not provide information about their employment status. About half (49%) of participants were married, while 6% were divorced, 2% were widowed, and 43% were unmarried.

Of the 266 pairs, 160 were monozygotic (MZ; 45 male–male, 115 female–female pairs) and 106 were dizygotic (DZ; 11 male–male, 49 female–female, 46 unlike-gender pairs).

Zygosity was assigned by a standard questionnaire regarding physical similarity of the twins during infancy; this is a well-established procedure in twin studies, which is known to be over 90% accurate. The reliability of this method in the ITR population was recently estimated in an independent sample of 158 same-gender adult twin pairs by using nine microsatellite markers; 149 pairs (94.3%) were correctly classified by the questionnaire.

Written informed consent was obtained from the twins, after complete description of the study.