

Monozygotic Twins Concordant for Female-to-Male Transsexualism: A Case Report

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Transsexualism is a rare phenotype with an estimated frequency varying between 1 in 10,000 up to 1 in 100,000 in male-to-female transsexualism (MTF) and 1 in 30,000 up to 1 in 400,000 in female-to-male transsexualism (FTM) (Cohen-Kettenis & Gooren, 1999). Various explanations have been suggested ranging from biological to psychosocial factors (Coates, 1990; Cohen-Kettenis & Gooren, 1999; Zucker & Bradley, 1995). Emerging evidence supports brain morphological, physical, and genetic differences in transsexual persons (Bosinski et al., 1997; Henningson et al., 2005; Kruiver et al., 2000; van Goozen, Slabbekoorn, Gooren, Sanders, & Cohen-Kettenis, 2002; Zhou, Hofman, Gooren, & Swaab, 1995). Nevertheless, little data exist supporting any single hypothesis. A few monozygotic (MZ) FTM and MTF twin pairs, concordant or discordant for transsexualism, have been reported (Segal, 2006). However, only a single MZ concordant FTM twin pair has been published (Sadeghi & Fakhrai, 2000). We report the second female MZ twin pair concordant for transsexualism.

The sisters (BK and RK) grew up in a rural environment in Macedonia. They have three older brothers and three older

sisters. Their father, a severe person, worked abroad and rarely saw his family (once every two years). The mother and an older brother assumed the parental roles for the girls. The mother was described as a “good mother” who “managed” the entire family. BK, the older twin sister, described herself as more reserved compared to the younger sister, RK, who described herself as livelier. BK was more frequently ill until the third year of life. RK reached her developmental milestones (walking and talking) around four months earlier than BK. RK had several accidents in childhood and adolescence; one accident was followed by a cerebral concussion. In contrast, the similarities between the girls were striking. Both sisters preferred to play with boys rather than girls since age six years. They tried to imitate all the things that boys did. They played soccer and other boys’ games. Both rejected dolls and girls’ clothing. Both twins preferred short hair and forced their mother to cut their hair short from age six years. In school, both sisters preferred sports, music, and history. At school, they were asked if they were boys or girls by the teachers and responded that they were boys. Both reported many conflicts with other children. With the onset of menarche and breast development, they began to use bandages to flatten their breasts. They had not been sexually educated by their parents and prayed each evening to become and grow up as boys. The sisters began hating their bodies and stopped playing soccer.

After the 8th grade, the twins moved to Germany to live with their father because of the growing social and family pressure related to their gender identity problems. The father forced his daughters to visit a school with mainly girls; however, both were treated as boys. Considerable conflicts with the father developed. After the twins were legally independent, the twins moved to Berlin and decided to undergo a sex-change operation at age 25 years. Both twins denied heterosexual or homosexual contact and both were not

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sexually attracted to either men or women at the time of examination.

At the time of assessment, the twins were aged 30 years and had begun taking male sex hormones six months earlier. They dressed as males. Physical examination revealed a normal external female phenotype. Both sisters still bandaged their breasts. Gynecological examination before hormone treatment revealed no abnormalities. Psychological testing with various questionnaires (Freiburger Personality Inventory; Beck Depression Inventory; State-Trait Anxiety Inventory) revealed a slightly elevated depressive tendency in both sisters. There was no indication of any additional psychiatric disorders in either the sisters or in first degree relatives. The diagnosis of transsexualism was based upon ICD-10 criteria and according to the “Health Law Standards of Care for Transsexualism” criteria.

Hormonal analysis revealed no abnormalities (testosterone, free testosterone, estradiol E2, sexual hormone binding globulin, FSH, LH). Cortisol and 17 α -hydroxyprogesterone before and after ACTH stimulation were in the expected range. Chromosomal analysis revealed a normal female karyotype (46, XX) in both sisters. Molecular genetic analysis using 15 microsatellite markers was performed to confirm monozygosity. The probability of monozygosity was calculated to be 99.9994%.

Several MZ twin pairs and one triplet (FTM and MTF) concordant or discordant for transsexualism have been reported (Anchersen, 1956; Buhrich, Bailey, & Martin, 1991; Garden & Rothery, 1992; Green, 2000; Green & Stoller, 1971; Hyde & Kenna, 1977; McKee, Roback, & Hollander, 1976; Sadeghi & Fakhrai, 2000; Segal, 2006; Zucker & Bradley, 1995). Only five pairs represent FTM MZ twins. Four pairs were discordant (Garden & Rothery, 1992; Green & Stoller, 1971; Segal, 2006) and only one was concordant for transsexualism (Sadeghi & Fakhrai, 2000). Seven additional MZ FTM transsexual twin pairs (two concordant and five discordant) were reported by Diamond and Hawk (2004).

Twin studies allow estimating the relative contribution of genetic and environmental factors to a specific phenotype (Boomsma, Busjahn, & Peltonen, 2002). For example, Coolidge, Thede, and Young (2002) used a child and adolescent twin sample with gender identity disorder and estimated an additive genetic component of 62% and a non-shared environmental component of 38%. These estimates were achieved by comparing the phenotypic correlations of MZ versus dizygotic (DZ) twin pairs. Concordant MZ twins only allow estimation of the genetic contribution, but do not allow the estimation of shared environmental factors as a second potential source for similarity. MZ twins discordant for a trait may shed light on relevant environmental influences as they share the same genetic background (Segal, 2006). Therefore, the relative influence of genetic and envi-

ronmental factors of transsexualism could only be estimated analyzing a set of MZ and DZ twins, optimally derived from a population-based registry. Such a registry would include data concerning twinning and transsexualism. The MZ twin sisters presented here described an almost identical development, including sexual identity development. Since they shared the same genetic and familial (environmental) background, they do not allow conclusions regarding genetic or environmental influences on the development of transsexualism. Because of the low number of twin pairs and the lack of published reports of DZ twins concordant or discordant for transsexualism, an estimation of the heritability of transsexualism awaits additional systematic studies. Therefore, we suggest that DZ twins should be reported in the future and a systematic collection of twins with FTM and MTF transsexualism should be initiated.

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