Identical Twins Raised as Sister and Brother

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Case Presentation

Birth: German parents, mother 28, father 32 years of age, healthy, no consanguinity. 1986 mother dies 11 yrs. later because of ovarian cancer. A friend of the mother is rearing the twins. The results of 18 blood parameters demonstrate, that they are identical twins both having a chromosomal gender of 46,XY. They are raised as female (Andrea ) and as male (Andreas ). Their medical gender assignment was chosen according to the appearance of their external genitalia. names were changed for subject protection.


Andreas (born 1985) ambiguous external genitalia (Sinnecker 2) microopenis, hypospadias, flat scrotum inguinal hernia, 2 gonades chromosomes: 46,XY gonadal biopsy: testicular tissue, no Sertoli cells? no ovarian structures

Introduction

A follow-up of identical twins with 46,XY chromosomes and DSD is presented. One of them was raised as a girl, the other one was raised as a boy according to the ambiguity of their genitalia, which were adjusted to female and male, respectively. Medical gender assignment was done without an exact diagnosis, puberty was induced by female and male hormones, which were substituted in adulthood, too. Sister and brother developed well mentally and physically. Both of them express their satisfaction with the former medical decisions.

Background

Disorder of sex development (DSD) can be caused by numerous hormonal and genetic defects, such as enzyme deficiencies in steroid production or a mutation in the SRY-gene, disturbing regular sex differentiation (Lukas-Herald). In our cases we assume a genetic defect at the SRY-gene causing a marked undervirilisation in the girl and a milder effect in the boy.

Figure 1: SRY (sex determining region Y protein = testis determining factor) structure and location on the short arm of the Y-chromosome

Summary and Conclusion:

1. An assumed mutation of the SRY-gene causes differently expressed disorders in gonadal development in these identical twins: Andrea's disorder is so pronounced, that no functional gonades (but only streak gonades) are developed, also no male structures inspite of 46,XY. Swyer's-syndrome (King)

2. The mutation of the SRY-gene in Andreas is disturbing the development of testes and of testosterone to a lesser degree, but blocks the production of AMH responsible for suppression of Mullerian ducts: in consequence a phallus is developed and the female structures remain: Hernia-uteri-syndrome / Oviduct persistence / Persistent Mullerian duct syndrome (PMDS) (Bastian; Johansen).

3. The medical assignment of gender was well tolerated. Both patients live in a stable social surrounding, both of them have an academic profession, both of them report of a good heterosexual partnership with a normal vita sexualis. Both of them wish to rear children with their partners.

4. They are very content with the gender assignment given in their infancy. They can not imagine to have lived somewhere inbetween such as in a third gender.

References


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Figure 2: growth chart of 46,XY-twins raised as a boy (+) and as a girl (-)

Figure 3: SRY (sex determining region Y protein = testis determining factor) structure and location on the short arm of the Y-chromosome

Poster presented at: