# 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 choosen according to the appearence of their external genitalia.
names were changed for subject protection.

**Andrea (born 1985)**

external genitalia: female (Sinnecker 5)

chromosomes: 46,XY

Androgen receptor studies neg.
(I.A. Hughes)

1889: explorative laparatomy:
streak gonades exstirpated, no ovary- or testis-specific structures; vagina, portio, uterus, Fallopian tubes.

1994: family moves from former „DDR“ to Hamburg, follow-up at the university;
repeated analysis of chromosomes: 46,XY in both of the twins;
repeated analysis of *AR*-genes (Hiort): no androgen-receptor-insensitivity;
PCR-amplification of *SRY* genes(Gal): no difference compared to male controls

2013: molecular analysis of *SRY*-gene refused by patients

1997: female puberty was induced by etrogens and gestagens

1998: cyclic treatment
LH: 24,7 FSH: 87,4 mU/ml
Estradiol: 51 pg/ml

2005:treatment interrupted by patient:
mood swings, bad feeling

2013:no medication:
LH: 17,2 FSH: 87,5 E2 < 5
→ Estradiolvalerat-gel
→ Vit.-D3

Adult height: 183 cm, weight 68 kg

**Andreas (born 1985)**

ambiguous external genitalia (Sinnecker 2)
micropenis, hypospadia, flat scotum
inguinal hernia, 2 gonades
chromosomes: 46,XY
gonadal biopsy: testicular tissue,
no Sertoli' cells ? no ovarian structures

1985: urethro-cystoscopy: female shaped urethra, vagina, portio

1986: explorative laparatomy:
testes with epididymis, vagina uterus, Fallopian tubes, no ovaries.
All structures exstirpated

1989: penile plastic operation

1990: urethra reconstruction

1993: artificial testes for scrotal enlargement

1998: male puberty was induced by Testosterone-Depot
LH: 19,2 FSH: 39,6 mU/ml
Testosterone: 12,7 ng/ml

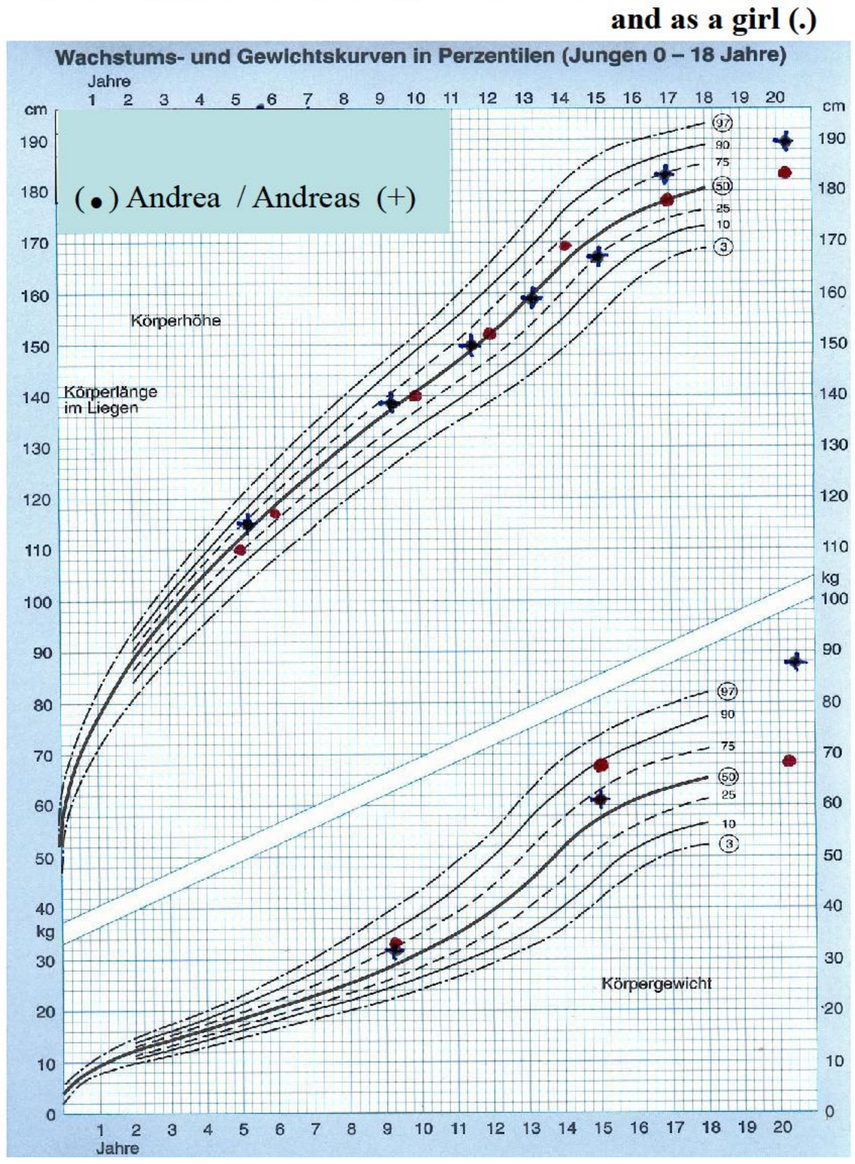
2008: Testosterone-Depot
LH: 19,7 FSH: 61,6 T: 1,9

2009: Nebido 1000 mg / 3 mon.

2013: LH: 11,3 FSH: 32,6 T: 4,1

Adult height: 190 cm, weight: 88 kg

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



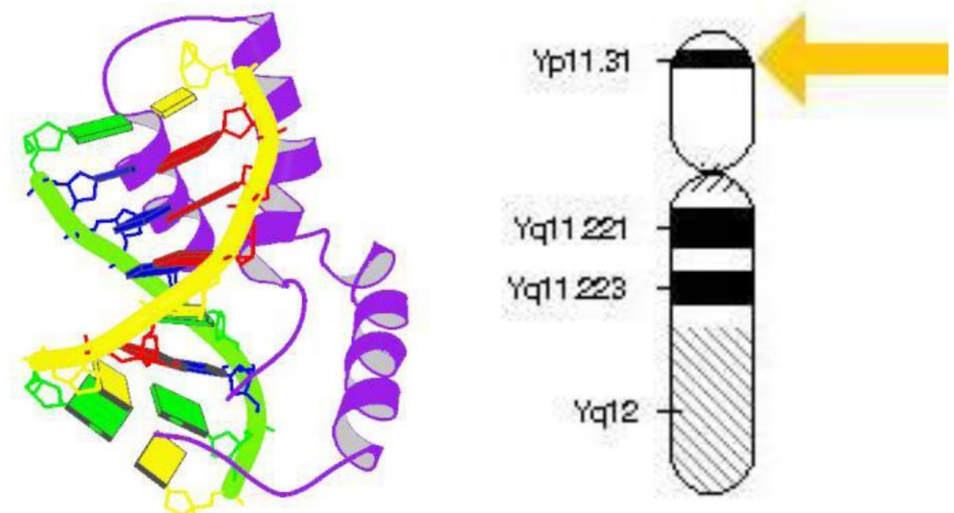

**Introduction**

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-Harald).
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 determing 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 Muellerian ducts: in consequence a phallus is developed and the female structures remain: Hernia-uteri-syndrome / Oviduct persistence / Persistent Muellerian duct syndrome (PMDS) (Bastian; Johansen).

3. The medical assignment of gender was well tolerated. Both patients live in a stable social sorrounding, 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.

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