

Society for Endocrinology, Clinical Update 2007

A Case of XY Sex Reversal

Shyam Kalavalapalli, A.Heald, H.Buckler
Salford Royal Hospitals NHS Trust, Salford

A girl presented to us at age sixteen with primary amenorrhoea, poor breast development and tall stature.

She is the youngest of the 11 children in a Jewish family. One of her brothers had ambiguous genitalia at birth requiring reconstructive surgery as did one of her sister's sons. Another sister's son had hypospadias. Her parents and family are not aware of her condition.

On first presentation she had marfanoid features with Tanner stage of Breast-1, Axillary hair-1 and Pubic hair 3. She is however phenotypically female with mild clitoromegaly.

Her endocrine profile confirmed primary gonadal failure with Oestradiol <73pmol/l, LH – 21.5 U/L, FSH – 62.7U/L, SHBG – 22 nmol/L, Testosterone: 1.8 nmol/L, F.A.I – 3.6, Prolactin – 315 with normal thyroid and adrenal axes.

Her karyotyping revealed 46XY. Her bone age was 13 with a chronological age of 18 years. Her most recent imaging showed a presumptive tiny uterus and ovoid gonadal structures on each side of the mid line. SRY FISH studies showed the presence of SRY gene and no mutations were identified on the SRY gene mutation studies.

We proceeded with HCG Stimulation test which failed to establish the presence of functioning testicular tissue with her basal testosterone of 1.8nmol/L rising only to 2nmol/L post stimulation. Her bone density suggested osteopenia with Z –scores of L1 – L4 spine: -1.7 and Left Hip: -1.2.

In conclusion, ours is a case of undervirilized XY due to XY gonadal dysgenesis. She is managed as a female with Climagest 2mg and was referred for breast augmentation surgery with a breast stage of 4. We also sought gynaecology opinion for possible laparoscopy and histological confirmation and resection if gonadal tissue is confirmed to be testicular in origin.