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SRY (Sex Determining Regions in Y) Basis of Sex Reversal in XY Females

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ABSTRACT 46,XY sex reversal condition is known to be caused by the point mutations or deletions in SRY gene at Yp11.3. Individuals with 46,XY status are phenotypically females with gonadal dysgenesis and poorly developed secondary sexual characters. In the present study, it is aimed to report the SRY basis of sex reversal in 25 XY female probands aged 4 to 39 years. The karyotypes in 21 were 46, XY and 45, X/46, XY in 4. The presence of the SRY gene was determined in 20 probands (80%), out of which 18 had 46, XY and 2 had 45, X/46, XY karyotypes. SRY gene was not detected in 5 probands (20%), among them 3 had 46,XY and 2 had 45,X/46,XY karyotypes and in these 5 case, SRY gene was considered to be deleted. The present study confirmed the importance of the genetic evaluation of the SRY gene in females with XY status for further management.