

# Challenges in managing 46, XY partial gonadal dysgenesis in Saudi Arabia



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### BACKGROUND

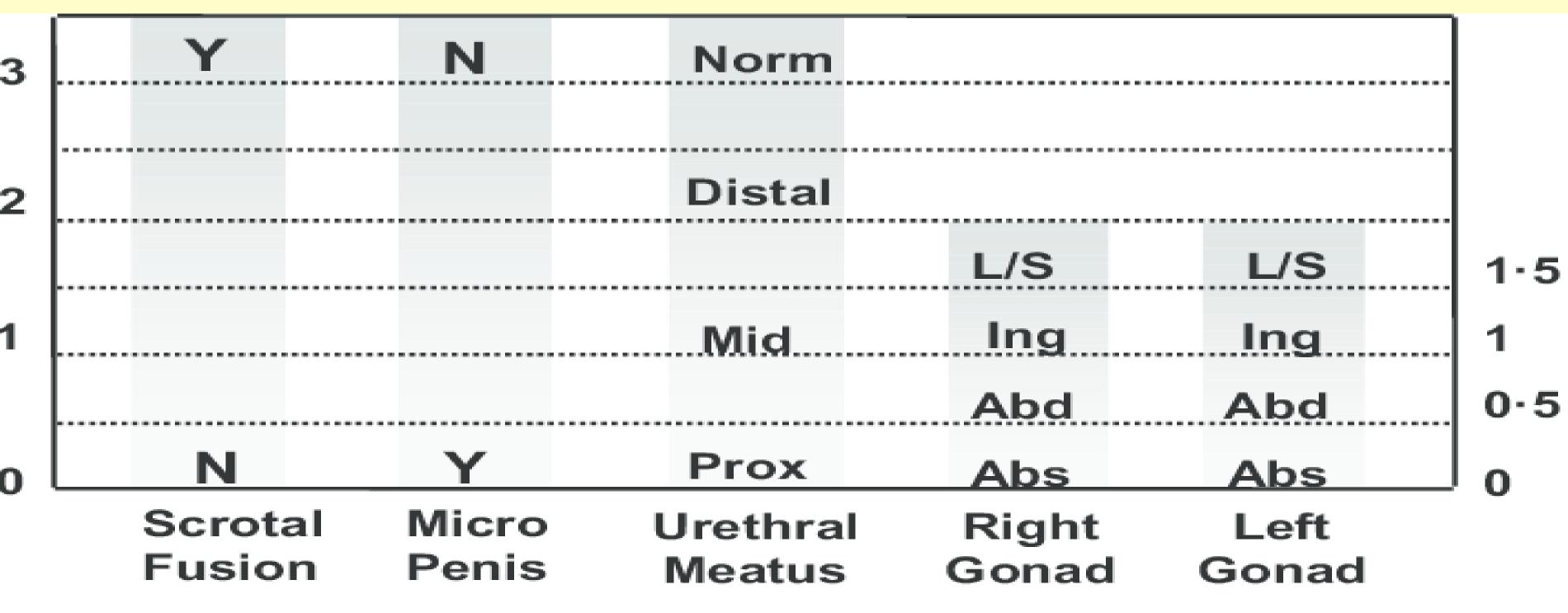
- Partial gonadal dysgenesis is a rare 46,
  XY Disorder of sex development (DSD).
- Many studies examined the challenges in presentation and gender assignment regarding the genital features, genetic mutations and histopathological risks of dysgenetic gonads.
- More recently some studies described the long-term outcome of patients reared as males.
- In the Middle East, cultural aspects might also influence the parents' choice of gender assignment.

## **CASE REPORTS**

- We report on a Saudi newborn with 46, XY chromosomes, presence of the "sex-determining region of Y chromosome" (SRY) gene, inguinal gonads, fallopian tubes, uterus, common urogenital sinus, and ambiguous genitalia with a micro phallus like structure and perineal hypospadias giving 8-9/12 of external masculinization score (EMS).
- Gonadal biopsy revealed virtually normal testicular tissue in both gonads. Her 1-year-old sister, born in district settings with limited resources, had a similar presentation at birth and was treated for labial adhesions in surgery clinics with unsuccessful attempts of dilatations. She is found to be a copy/paste of the baby sister clinically and genetically..



USS and MRI showed testes and no ovaries. Bilateral inguinal testes confirmed by biopsy



#### DISCUSSION

Parents were extremely anxious of gender reassignment in the older sister. Psychosocial and cultural pressures heavily influence parental decision making in this neck of the wood. XY DSD girls would potentially have a very gloomy future of sexual life. This would support raising these patients as males in such communities but not necessarily gender reassignment that could be more stressful to parents. However, a trial of testosterone therapy proving response of peripheral tissue and to avoid a concomitant pathology of androgen insensitivity is worth prior to final assignment if possible. Although, consideration of long-term outcomes in children with these disorders mainly affect the decision of gender assignment and reassignment, however, psychosocial pressures on parents also have a major role in this perspective. Removal of the gonads during surgery anticipating development of gonadal tumors has to be taken into consideration. While in the past, female sex assignment was commonly recommended for such patients, raising them in a male gender role is now more considered.

## CONCLUSION

Parents should be involved in the decision that is ultimately based on extensive analysis of the individual case. Extensive counseling and support to parents help them to take a decision in the best interest of the patients despite all of psychosocial pressures.







