PENILE AGENESIS- ADOLESCENCE PRESENTATION

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Case Report

A 14-year-old boy was brought by his parents with history of absent penis since birth. Family history was negative for congenital or familial diseases. He had been voiding through the anus and was continent. Examination revealed normally built boy with an absent penis, well-developed scrotum with bilateral normally descended testes (Fig-1). On per rectal examination the urinary meatus was felt as a dimple just within the anal verge. No other systemic anomalies were detected.

Investigations revealed a 46 XY karyotype, buccal smear negative for chromatin and normal renal function. The excretory urogram (IVU) showed bilateral normally functioning kidneys, normal ureters and bladder. Voiding cystogram revealed normal bladder, no reflux and drainage of contrast from the urethra into the rectum (Fig-2). The patient was advised gender reassignment or staged phallic reconstruction to overcome psychosociological embarrassment but he did not agree to either. Psychological evaluation revealed being well adjusted with his disability.

Discussion

Penile agenesis, albeit an exceedingly rare disorder with an occurrence estimated at 1 in 10 million births [1], about 70 cases have been reported in neonates. In most cases the diagnosis is not in question. Although the genetic sex in majority of aphallic patients is 46 XY and fertility theoretically may be possible, these

patients should be raised as girls [2]. The poor psychological outcome in patients who were raised as boys strongly reinforces this concept. The surgical difficulties and poor result encountered in phallic reconstruction have prompted female sex assignment [3]. Early sex reassignment in the neonatal period is ideal as gender awareness begins around 18 months after birth and is well-established in most children by the time they are 2.5 years old. Gender reassignment at adoles-

Fig. 1 : Clinical photograph showing absence of penis , well developed scrotum with normally descended testes.

Fig. 2 : Post IVU voiding cystogram lateral view showing contrast in urinary bladder (B) and rectum (R)

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cence is fraught with significant social and psychopathological trauma and may not be agreed to by the patients as in our case.

In addition, sex reassignment in the neonatal period with early orchidectomy may have the advantage of preventing the postnatal testosterone surge and central nervous system testosterone imprinting which are important to subsequent gender identity [3].

References

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