LETTER TO EDITOR (VIEWER'S CHOICE)

FAMILIAL HYPERCHOLESTEROLEMIA IN SIBLINGS WITH DIABETES MELLITUS

Shashidhar A, Bijal Rughani, S R Keshavamurthy

The infant weighing 3 kg, was born at term, by vaginal route, to a 26yr old non-consanguineously married gestational diabetic who required insulin for glycemic control. She had a previous normal childbirth and had no significant antenatal problems. The antenatal ultrasound had missed the anomaly and the child was noted to have complete inversion of external genitalia at birth. The scrotum was bifid. Both the testes were palpable in the well formed scrotal sac. The rudimentary penis was placed just anterior to the anal opening which itself was displaced anteriorly. (Figure 1) There was no hypospadias or chordee. A diagnosis of complete form of penoscrotal transposition with ectopic anus was made and the child was evaluated for other associated anomalies. The child also had a ventricular septal defect and atrial septal defect with a normal male XY karyotype while other investigations like cranial and abdominal ultrasonogram, thyroid profile, serum testosterone levels were normal. The baby underwent staged surgery at our hospital for the correction of the anomaly and is on follow-up.

Figure 1: Complete penoscrotal transposition

Complete penoscrotal transposition (CPST) is a rare and unusual malformation in which the scrotum is located cephalad to the penis, frequently associated with major and often life-threatening malformations involving the genitourinary, cardiovascular, or skeletal systems. (1) Parida et al (2) have noted a 90% incidence of major renal anomalies including complete agenesis of the urinary system, unilateral or bilateral renal agenesis, horse-shoe kidney, ectopic pelvic kidney, polycystic or dysplastic kidneys, obstructive uropathy with reflux, megaureter, and hydronephrosis. The most common non-urogenital abnormalities associated with PST were: mental retardation (60%), anorectal malformations (33%), central nervous system anomalies (29%), vertebral defects (29%), preaxial limb defects including radial dysgenesis (24%), and congenital heart disease (19%). It therefore appears likely that CPST may represent only one of the results of a major localized or generalized embryological insult to the fetus during the 4th-6th weeks of gestation, when the major organ systems are passing through a crucial phase of development and differentiation. Abnormal positioning of the genital tubercle in relation to the scrotal swellings during the critical period at 4-5 weeks' gestation may affect the inferomedial migration and fusion of the scrotal swellings. If the phallic tubercle is also intrinsically abnormal, development of the corporal bodies and the urethral groove and folds may be affected; this explains the frequent occurrence of the other genital abnormalities. In presence of normal penis this anomaly does not cause any sexual dysfunction. However, its surgical repair is for psychological reasons. (1)

REFERENCES

1. Chadha R, Mann V, Sharma A, Bagga D. Complete penoscrotal transposition and associated malformations. Pediatr Surg Int 1999; 15: 505-507
2. Parida SK, Hall BD, Barton L, Fujimoto A. Penoscrotal transposition and associated anomalies: report of five new cases and review of the literature. Am J Med Genet. 1995; 59: 68-75

From: Department of Pediatrics, Kempegowda Institute of Medical sciences, Bangalore, India.

Address for Correspondence: Dr Shashidhar, #114 KIMS Mens Hostel, BSK, 2nd Stage, Bangalore 70, India. Email: shashiishere@gmail.com

E-published: 1st February 2011. **Art**#13