children (n = 18)		
Clinical data and causes	No.	%
Clinical data		
Micropenis	16	88.9
Undescended testes (both)	9	50.0
Extragenital anomaly	7	38.9
Hypospadias	4	22.2
Undescended testis (single)	2	11.1
Death	2	11.1
Causes		
Generalized developmental malformation, with		

testosterone biosynthetic defect

Syndromic: Goldenhar syndrome

VATER association

biosynthetic defect

Potter syndrome

normal testosterone synthetic pathway

Generalized developmental malformation, with

Congenital local genital defect, with testosterone

Congenital local genital defects, with intact

testosterone synthetic pathway Androgen insensitivity syndrome 22.2

11.1

5.6

5.6

5.6

278

27.8

5.6

Table 2 Clinical data and likely causes of ambiguous genitalia in 46,XY karyotype