47,XXY With Associated Bilateral Renal Agenesis

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• We describe the case of a 36-week gestational-age male stillborn with bilateral renal agenesis and a 47,XXY karyotype, as well as features of Potter sequence. No other congenital abnormalities were noted. Severe oligohydramnios was diagnosed prenatally at 30 weeks, and cytogenetic analysis was performed postmortem. Urinary tract anomalies are uncommon in association with Klinefelter syndrome. Unilateral renal agenesis has been described. We describe, to our knowledge, the first case of bilateral renal agenesis in association with 47,XXY.

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Severe third-trimester oligohydramnios implies a higher risk for abnormal fetal karyotype, and it has been associated with fetal urinary tract anomalies in 8% to 33% of cases.[1]

Up to 15% of congenital urogenital anomalies occur secondary to an underlying chromosomal disorder. The incidence ranges from 60% to 100% in certain chromosomal aberrations, including trisomy D and E as well as Turner syndrome.[2,3]

Multiple congenital abnormalities have been associated with Klinefelter syndrome (47,XXY).[4] However, gross malformations of the urinary tract are rare.[2]

This article reports a case of bilateral renal agenesis in a fetus with 47,XXY. To our knowledge, this is the first such report.

REPORT OF A CASE

The propositus was a 36-week gestational-age stillborn, product of the second pregnancy of a 20-year-old healthy mother. The parents were nonconsanguineous. The pregnancy was complicated by severe oligohydramnios diagnosed at 30 weeks. Further testing, including magnetic resonance imaging at 31 weeks, showed bilateral absence of kidneys.

Postmortem examination showed a 1840-g stillborn male fetus with features of Potter sequence: compression deformity of the face, marked bilateral pulmonary hypoplasia, and talipes calcaneus varus. Bilateral renal agenesis was confirmed (Figure 1). The ureters were absent bilaterally. The bladder was tubular and hypoplastic. No other congenital anomalies were found.

A sample from the axillary skin was submitted for chromosomal analysis, which was performed by the high-resolution G-banding technique.

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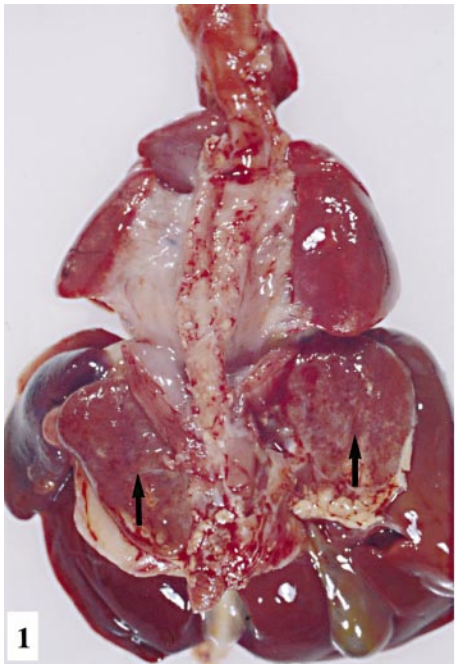

Figure 1. *En bloc dissection, posterior view. Note bilateral absence of kidneys and downward placement of round adrenal glands (arrows).*

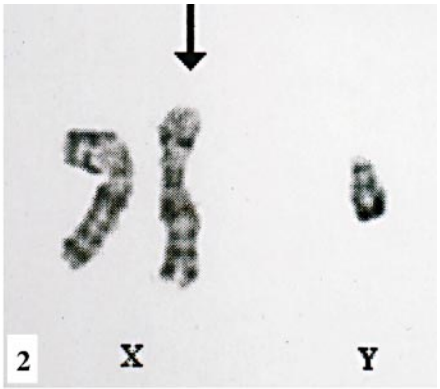

Figure 2. *47,XXY karyotype. Note presence of duplicated X chromosome (arrow).*

Examination of 3 metaphases revealed an abnormal male karyotype, 47,XXY, in all 3 (Figure 2).

COMMENT

Bilateral renal agenesis has been suggested to be multifactorial[5–8] and has also been associated with the expression of a single dominant gene. Up to 10% of all newborns have significant abnormalities of the urinary system. The incidence of bilateral renal agenesis is 0.3 to 0.4 per 1000 newborns but reaches 3 per 1000 in stillborn fetuses.[2]

Malformations of the urinary tract do not seem to occur more frequently in individuals affected with Klinefelter syndrome than in the normal population.[2] Although uncommon, certain abnormalities of the urogenital tract have been described in association with the 47,XXY karyotype, including renal cysts,[2] atresia of the pyelic cavity,[9] atrophic scrotum, rudimentary penis,[10] cryptorchidism, hypospadias, and unilateral renal agenesis and deformities.[4] The case we describe appears to be the first case of 47,XXY with associated bilateral renal agenesis.

Fetuses with Klinefelter syndrome are not likely to be dysmorphic; therefore, cytogenetic analysis might not be performed at the time of postmortem examination in stillborn fetuses with seemingly isolated bilateral renal agenesis. For this reason, it is possible that previous cases of bilateral renal agenesis in fetuses with 47,XXY have been overlooked.

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