

# Prenatal management of inherited urogenital malformation: Case report

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## Summary

Documentation of unique kidney renal function early in pregnancy can be helpful in defining prenatal management and therefore in improving prognosis. Antenatal diagnosis of a solitary kidney was performed at 20 weeks' gestation in a foetus with a 1,7 chromosome translocation. Because of the decreasing renal function and the increasing pelvic dilatation, an early in utero stenting was placed at 23 weeks' gestation. Optimal outcome occurred and the baby was delivered at 32 weeks. Complete assessment of the malformation showed a left hydronephrosis due to a megaureter, right renal agenesis with ipsilateral cryptorchidism and agenesis of the right vas deferens. The chromosomal translocation was inherited from the mother who was affected by uterus didelphys, obstructed right hemivagina and right renal agenesis.

Renal function of the unique kidney with hydronephrosis can be early diagnosed and promptly treated. This condition should also increase the index of suspicion of underlying genital and chromosomal anomalies.

**Key words:** Renal agenesis; Urogenital anomalies; Abnormalities vas deferens.

## Introduction

Renal agenesis is frequently associated with genital anomalies and urinary tract obstruction of the contralateral kidney. Prenatal discovery of this condition requires prompt clinical evaluation and renal function testing in order to predict fetal prognosis [1, 2]. In utero intervention is often mandatory to decompress urinary tract obstruction and reestablish amniotic fluid volume. We describe a case with inherited urogenital malformation, obstructive uropathy and a chromosomal 1,7 translocation.

This report emphasizes the necessity of early prenatal diagnosis and treatment of the obstructive uropathy in a solitary kidney. A minimally invasive procedure could be life-saving in early pregnancy.

## Case report

The mother was a 25-year-old primipara. Before pregnancy she underwent clinical and radiological investigations for infertility. She was found to have uterus didelphys with a right cervical mass with obstructing vaginal septum and ipsilateral renal agenesis. Excision of the vaginal septum and marsupialisation of the blind vagina was performed before pregnancy.

During pregnancy, renal fetal abnormalities were suspected at 20 weeks of gestation. Serial sonographic investigation showed a left obstructive uropathy, right kidney agenesis and a small bladder. The fetus was implanted in the left uterus.

Ultrasonography at 23 weeks' revealed a severe grade 3 obstructive uropathy. Initial signs of renal failure were demonstrated by fetal urinary electrolyte measurements. A pyeloamniotic shunt was performed at 24 weeks' which resulted in a significant decrease in pyelic dilatation and improvement in renal function. Both parents were apparently healthy; they were

referred for genetic counseling and the karyotype of the mother showed a reciprocal balanced translocation 1, 7 (p22; p15). A cytogenetic study of the amniotic cells revealed the same chromosomal translocation of the mother.

The baby was delivered at 32 weeks' by cesarian section following premature labour due to maternal eclampsia. Birth weight was 1,800 g. The shunt was well positioned, with no abdominal wall disruption or migration.

Postnatal ultrasound, pyelography, micturition cystography and renal scintigraphy confirmed the left primary obstructive non refluxing megaureter and right renal agenesis with normal renal function. Bilateral inguinal hernias and right cryptorchidism were also discovered. The patient was managed by observation while the pyeloamniotic shunt remained in place for three months after birth. The shunt was removed after six months and the diuretic renography profile was found to have been significantly impaired since the birth. Bilateral inguinal hernia repair was performed at the age of 40 days: agenesis of the right vas deferens was discovered intraoperatively, the left vas deferens being normal.

## Discussion

Unilateral renal agenesis is reported in 0.1% of the general population; in 90% of the cases it is associated with ureteral agenesis and vesical hemitrigone. It is more common in males than in females, and it is more frequently left-sided. Absence of a kidney coexists in 70% of male patients with genital abnormalities. These include conditions such as agenesis of the vas deferens, ipsilateral cryptorchidism, cystic dysplasia of the testes and seminal vesical cysts [3, 4]. Cardiovascular, intestinal and bone disorders, cystic fibrosis and inguinal hernias have also been reported as extragenital anomalies [5, 6].

In females this malformation is combined with Mullerian duplication in a more complex urogenital malformation characterised by the syndrome of uterus didelphys,

obstructed hemivagina with renal agenesis and absence of ipsilateral tube and ovary. The association of congenital anomalies of the genital and urinary tracts is generally explained by the fact that both systems originate from a common urogenital ridge of the mesoderm. During development they differentiate so that, ultimately, any one of the terminal structures may be malformed.

The frequency of renal agenesis is also increased in patients with cervical, thoracic and sacral abnormalities such as the MURCS association, consisting of mullarian duct aplasia, renal aplasia and cervico-thoracic somite dysplasia [6].

Unilateral or bilateral renal agenesis and Mullerian anomalies, such as vaginal atresia or minor anomalies are often reported in hereditary renal adysplasia (HRA) which is pathogenetically related to an autosomal dominant trait inheritance, incomplete penetrance and variable expressivity [7].

Isolated renal agenesis has also been attributed to chromosomal defects with deletions on chromosomes 1, 6, 10, 13, 18, 21, X. A locus in a narrow region between 12q23 and 12q24 chromosomes is also known to be involved in the familial Darier Syndrome including keratosis follicularis, lumbar scoliosis and renal agenesis [6, 8].

Other similar entities have been described in the literature. There exists, for example, a "hand-foot-uterus syndrome", an autosomal dominant disorder combining limb and foot anomalies, defects of Mullerian duct fusion and urinary tract malformation in females and hypospadias of variable severity in males [9].

In our report the mother and her male offspring were both affected and both were carriers of an apparently balanced reciprocal autosomal translocation with breakpoints at 1q22 and 7q15: karyotype 46, XX, XY, t(1, 7) (p22; p15) respectively. This observation may suggest that the gene involved in the development of Mullerian and Wolfian ducts is probably located at these breakpoints [10, 11].

The antenatal discovery of a congenital solitary kidney should alert the physician to other associated pathologies because in 50% of cases there is also urinary tract obstruction. Prenatal detection of pelvic-ileoceleal distention with persistent hydronephrosis and cortical renal thinning of the unique kidney may have practical implications for the "in utero" management during the first half of pregnancy [11, 13].

## Conclusion

Renal agenesis associated with persistent obstructive uropathy and genital anomalies in the presence of a defect on chromosomes 1 and 7 is a complex malformation never previously described. A prompt in utero stenosis could improve prognosis by preventing renal failure.

This prenatal procedure can be life-saving if performed in the first half of pregnancy.

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