

# Concordant Partial Urorectal Septum Malformation Sequence in monozygotic twins

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**The partial urorectal septum malformation (URSM) sequence** is defined as a single perineal/anal opening that drains a common cloaca in combination with an absent (imperforate) anus. Internal pelvic structures typically show a cloaca with the bladder and rectum (and vagina in females) coalescing into a common canal that connect to the external surface in the perineal or anal area. The partial URSM sequence is associated with a variety of unusual external genital malformations and urinary tract anomalies. Abnormalities of the internal genitalia are common, with females having a bifid or septate vagina and bifid or bicornuate uterus (Müllerian duct defects).

We report a case of the partial URSM sequence in monozygotic twins concordant for this anomaly. Monozygotic twinning with this condition has not been previously reported. The diagnosis was confirmed postnatally and the babies underwent corrective urogenital and intestinal surgeries.

By definition, the partial URSM sequence is a milder expression of **the full URSM sequence**, which is defined as having no perineal or anal openings and is typically associated with an internal cloaca. It is important to differentiate the partial from the full URSM sequence because the prognosis in the partial URSM sequence is generally good, with long-term survival common. The URSM spectrum, which encompasses the partial and full URSM sequences, is believed to be caused by abnormalities of septation of the primitive cloaca.

## CLINICAL REPORT

The mother, a 19-year-old caucasian woman, gravida 1, para 0, presented to the Ultrasound Unit of The Department of Medical Genetic and Fetal Medicine at 18 weeks of spontaneous pregnancy for routine screening. There was no family history of congenital malformations. On sonographic examination, a monozygotic, diamniotic twin pregnancy was diagnosed. Twin A had dilated distal bowel loops with enterolithiasis (an ultrasonographic examination revealed an echogenic bowel with multiple foci of calcified meconium intraluminal). Twin B had dilated distal bowel loops. A possible diagnosis of anal atresia was made. The genitalia could not be clearly determined. In view of the abnormal sonographic findings, amniocentesis was performed. Both fetuses had a normal 46,XY karyotype. The patient was referred to the high-risk clinic. Fetal biometry was appropriate for gestational age and a normal amount of amniotic fluid was observed. To further rule out anorectal malformation, magnetic resonance imaging (MRI) was performed and demonstrated dilated distal bowel loops (with enterolithiasis in twin A). The parenchyma of the kidneys and urinary bladder appeared normal. Pregnancy was terminated by Caesarean (Cesarian - US) section at 35 weeks of gestation after premature rupture of membranes. Twin A was a male, weighing 1950 g. Twin B weighed 2350 g. Post-partum examination revealed close placental insertions of both umbilical cords, each containing three vessels. Over a length of 25 cm the umbilical cords, separated by amniotic membranes, ran in such close proximity that they appeared to have a common course. The monozygotic, diamniotic twin pregnancy was certified and concordant fetal abnormalities were diagnosed. Both neonates had a single perineal opening that drained a common cloaca in combination with anal atresia. The cloaca drained the bladder and colon separately. There was a short and hypoplastic colon, dilated distal bowel loops (in the twin A with enterolithiasis), and a fistula between the colon and the bladder. External genital malformations included: cloaca with a single opening, perineal hypospadias, bifid scrotum, and penoscrotal transposition. There were also pelvic and sacral abnormalities. The diagnosis of partial URSM sequence was confirmed postnatally and the babies underwent corrective urogenital and intestinal surgeries. At present they are 2 years old and their health condition is generally good.

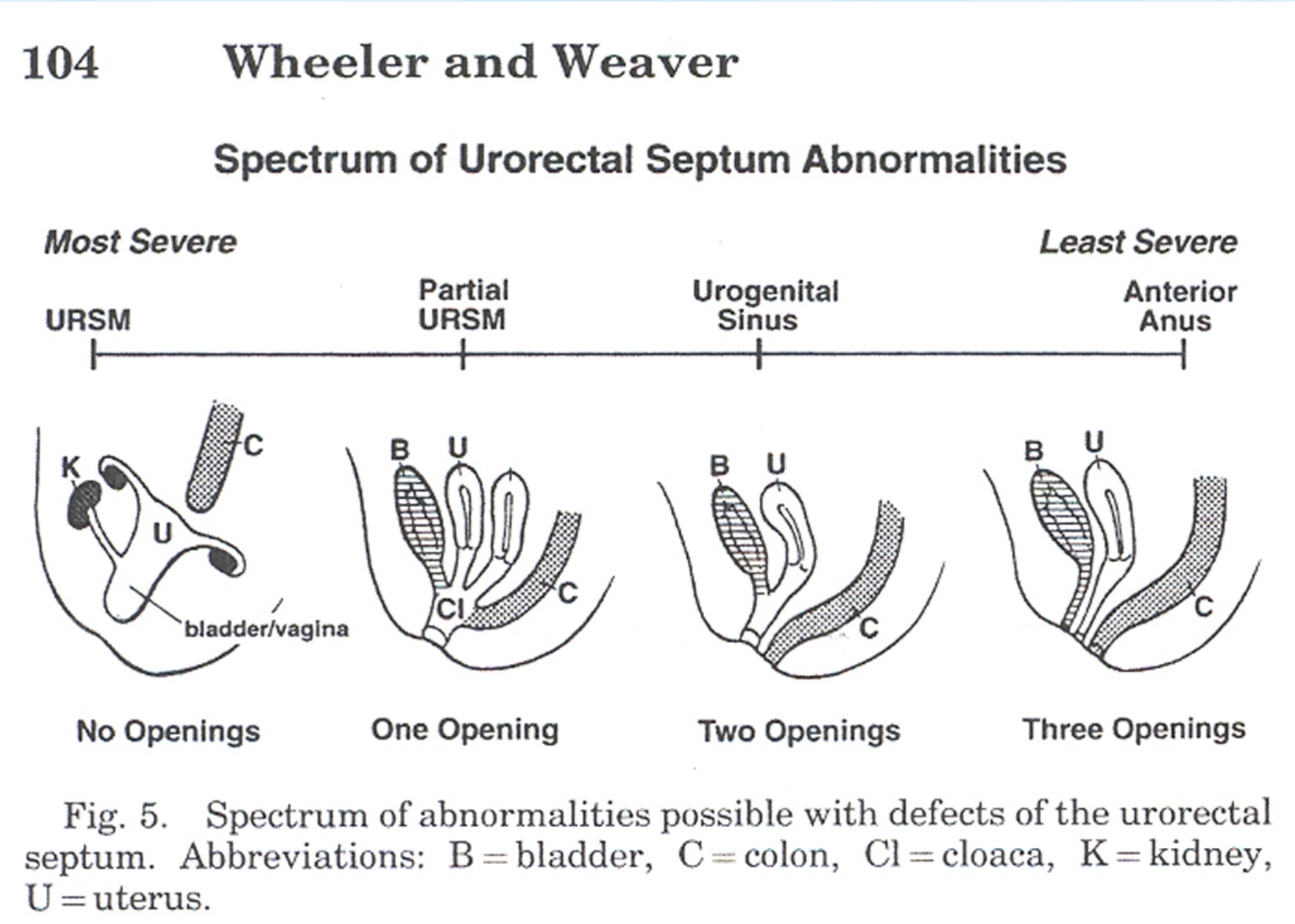


Fig. 5. Spectrum of abnormalities possible with defects of the urorectal septum. Abbreviations: B = bladder, C = colon, Cl = cloaca, K = kidney, U = uterus.

Wheeler PG, Weaver DD.  
Am J Med Genet 2001; 103: 99-105.

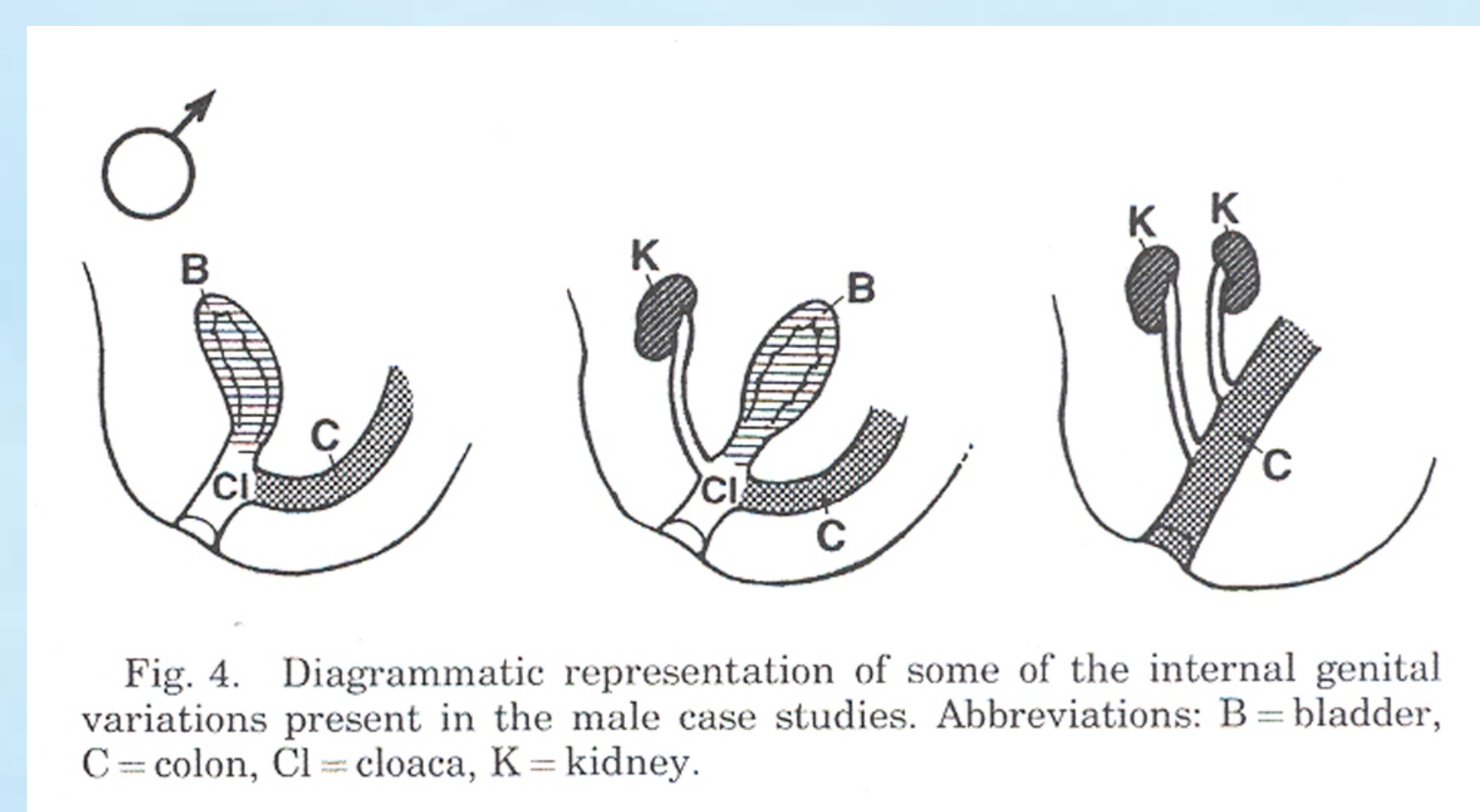


Fig. 4. Diagrammatic representation of some of the internal genital variations present in the male case studies. Abbreviations: B = bladder, C = colon, Cl = cloaca, K = kidney.

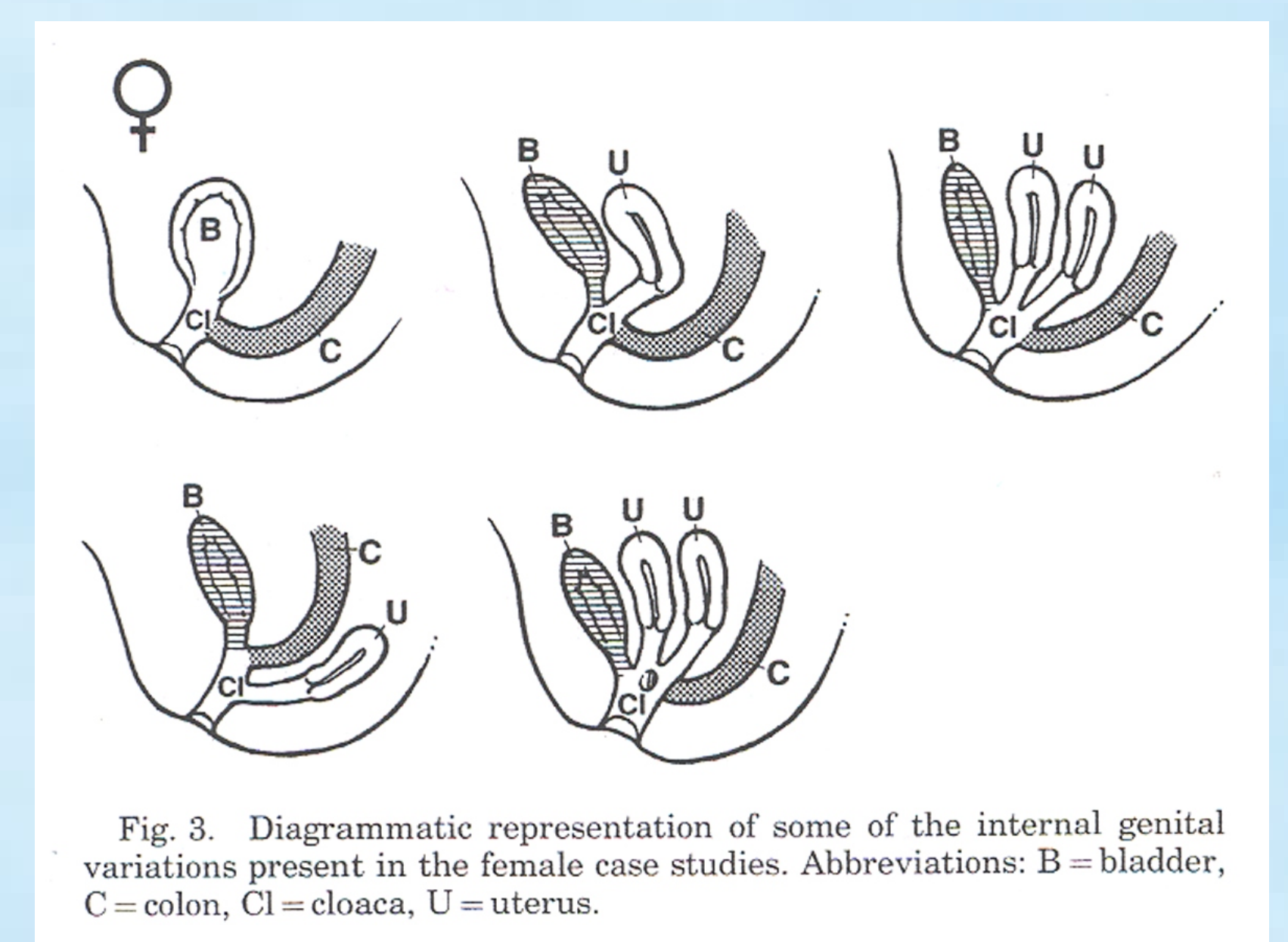


Fig. 3. Diagrammatic representation of some of the internal genital variations present in the female case studies. Abbreviations: B = bladder, C = colon, Cl = cloaca, U = uterus.

**Figure 1.** External genitalia of the twin A with cloaca single opening, perineal hypospadias, bifid scrotum and penoscrotal transposition.



**Figure 2.** External genitalia of the twin B with cloaca single opening, perineal hypospadias, bifid scrotum and penoscrotal transposition.



**Figure 3.** Monozygotic placenta with common placental insertion of umbilical cords and their common course in length 25 cm.

