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BRIEF COMMUNICATION

First Trimester Diagnosis of Cloacal Exstrophy: A Discordant Anomaly in Monochorionic Monoamniotic (MCMA) Twins—Case Report

Nupur Shah[1]

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Abstract Cloacal exstrophy is a rare anomaly, with a severe disruption of the urogenital tract resulting in a central mass of exstrophic bowel field flanked by two hemibladders. The reported case highlights the diagnosis of this discordant anomaly in monochorionic monoanmniotic twins as early as 12 weeks. It underscores the major ultrasound criteria like non visualisation of fetal bladder and infra-umbilical protruberant heterogenous mass which have an important role in early prenatal diagnosis of the anomaly.

Keywords Prenatal · First trimester · Cloacal exstrophy · Ultrasound

Case Report

The reported case is that of spontaneously conceived, 12 week gestation, monochorionic Monoamniotic (MCMA) twin pregnancy in a 34 year old woman. On the ultrasound, both the twins were in the same sac with no membrane separating them and a common placenta, suggestive of MCMA chorioamniocity. As shown in Fig. 1, for one of the twins, the urinary bladder was not visualised in its usual location and there was an infra-umbilical, well defined, protruding mass of heterogenous echogenicity. The kidneys appeared normal for gestation. The umbilical cord showed single umbilical artery outlining the mass on one side only. The thighs were placed wide apart. The skin over the spine was intact. The external genitalia was not visible for this twin. The co-twin had male external genitalia with no obvious structural defects. On ultrasound, the provisional diagnosis made was MCMA twins discordant with anomaly of cloacal exstrophy and single umbilical artery.

The parents refused any prenatal invasive procedure for fetal karyotype or for selective reduction of anomalous twin with radiofrequency ablation/interstitial laser [1]. They decided to terminate the entire pregnancy. After termination, abortuses were evaluated as shown in Fig. 2. Final diagnosis was confirmed to be cloacal exstrophy. Affected abortus' karyotype was done and was reported normal. The other abortus appeared normal structurally.

Discussion

Cloacal extrophy is a severe disruption of the urogenital tract resulting in a central mass of exstrophic bowel field flanked by two hemi-bladders (each having a urethral orifice). It occurs in 1 in 2,50,000 live births [2–4]. Embryologically, cloacal exstrophy has been attributed to premature rupture of the cloacal membrane; however, multiple alternative theories exist, such as abnormal over development of the lower cloaca that prevents mesenchymal tissue migration [4].

Cloacal exstrophy can be diagnosed as early as first trimester on prenatal ultrasound as in this case. Non visualisation of bladder on prenatal ultrasound was proposed to be important clue to bladder or cloacal exstrophy [5–7]. Austin et al. has proposed major and minor criteria for establishing prenatal diagnosis of cloacal exstrophy: major ultrasound criteria for diagnosing cloacal exstrophy prenatally are non-visualization of the bladder, a large midline infraumbilical anterior wall defect or cystic anterior wall

✉ Nupur Shah
nupurmshah@gmail.com

[1] Paras Bliss Fetal Medicine, Chandigarh, Haryana, India

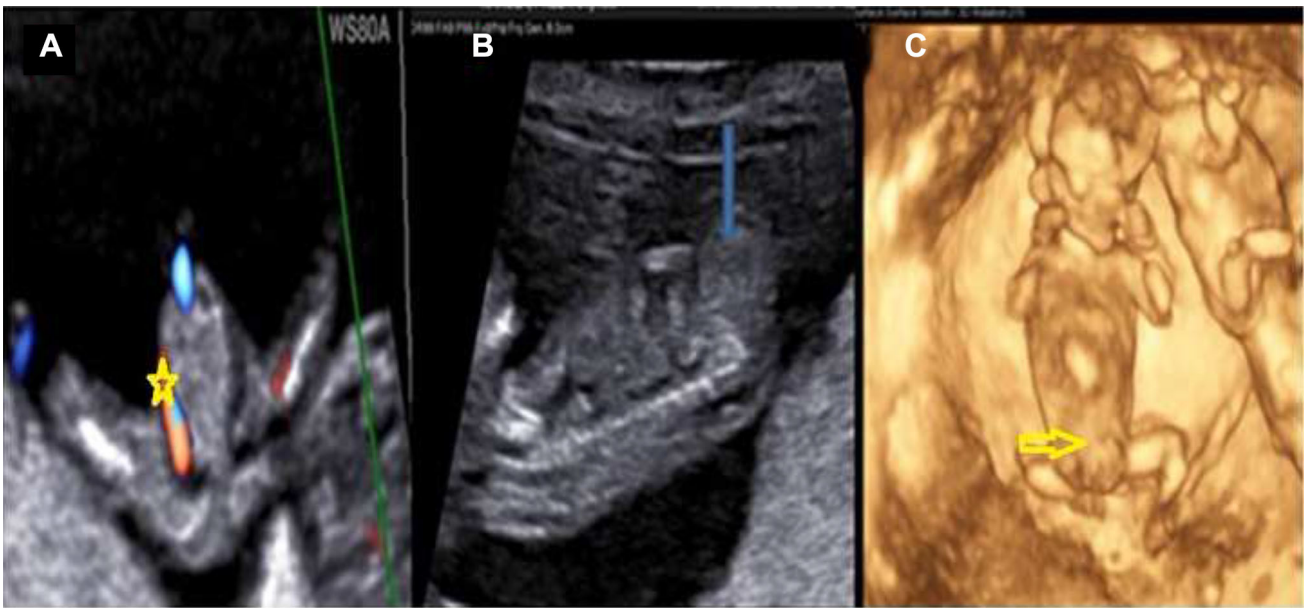

Fig. 1 Ultrasound appearance of cloacal exstropy. **a** Axial section suggestive of absent bladder, single umbilical artery (asterisk) outlining around heterogenous mass and wide apart thighs. **b** Saggital section suggestive of lower abdominal infraumbilical heterogenous mass (arrow). **c** 3D image suggestive of MCMA twins with infraumbilical protruding mass in one twin (yellow arrow)

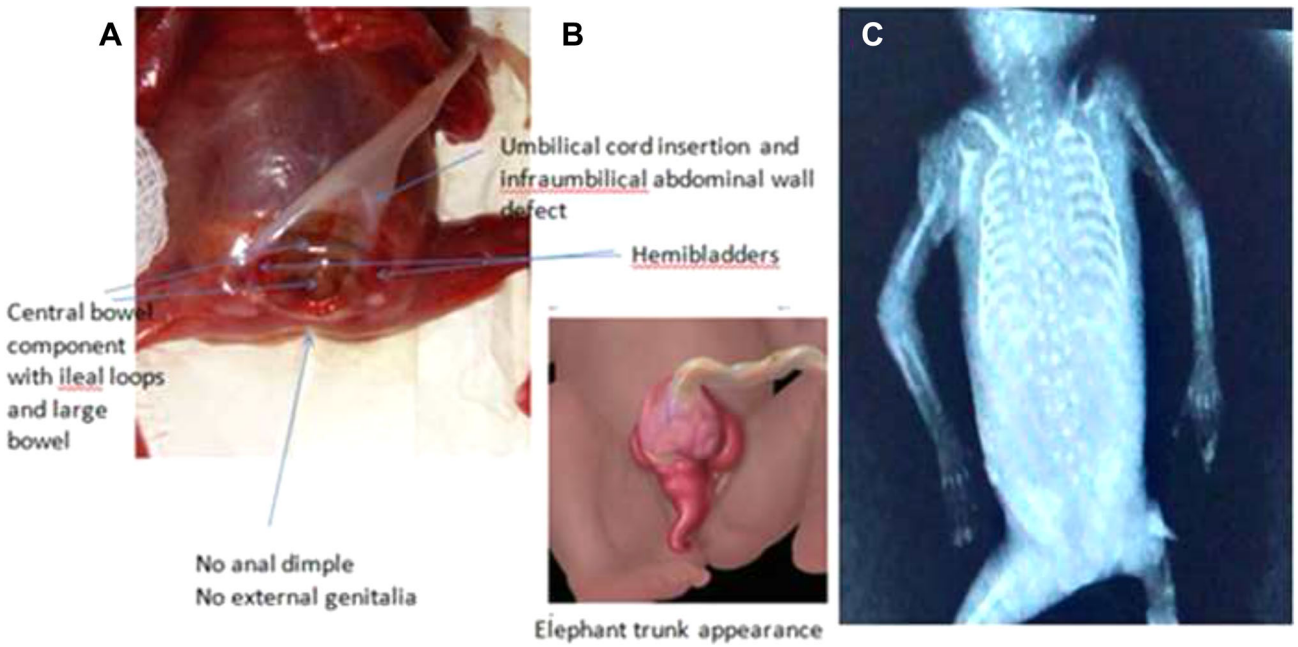

Fig. 2 Clinical picture and X ray findings of abortus. **a** Abortus showing infraumbilical mass covered with membrane, consisting of central bowel component and laterally shelved by hemibladders, no anal dimple and external genitalia seen. **b** Elephant trunk appearance. **c** X-ray of abortus showing normal spine, no splaying of vertebrae

structure (persistent cloacal membrane), omphalocele and lumbosacral anomalies. Seven less frequent or minor criteria include lower extremity defects, renal anomalies, ascites, widened pubic arches, a narrow thorax, hydrocephalus and single umbilical artery [6]. In our case, the diagnostic criteria like non visualisation of fetal bladder, infraumbilical protruberant mass, wide apart thighs and single umbilical artery give an important clue to cloacal exstrophy. However, the external genitalia and anal dimple cannot always be identified by ultrasound in first trimester. Differential diagnosis of cloacal exstrophy are bladder exstrophy, omphalocele, gastroschisis and complex

anterior abdominal wall defects. It can be associated with other anomalies e.g. OEIS complex (omphaocele, exstrophy, Imperforate anus and spinal defects) and Bladder Exstrophy–Epispadias–Cloacal Exstrophy, which warrant comprehensive anomaly scan [4, 5]. Cloacal exstrophy is not usually associated with chromosomal abnormalities, though some case reports noted 1p36 deletion especially with OEIS complex [8]. After ascertaining the ultrasound diagnosis, parental counselling in terms of prognosis, need of postnatal surgery and issues of incontinence should be discussed in association with paediatric surgeon. Before the age of viability of the fetus, termination of pregnancy can be offered.

Conclusion

First trimester diagnosis of cloacal exstrophy is possible on ultrasound using the above criteria, most important being infra-umbilical lower abdominal mass and non-visualisation of bladder. Comprehensive anomaly scan and appropriate counselling are warranted to decide further management of pregnancy.

Compliance with Ethical Standards

Conflict of interest The author declares that she have no conflict of interest.

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