Nigerian Journal of Perinatal and Neonatal Care

CASE REPORT

Macrosomic Infant of a Diabetic Mother with Hydrometrocolpos Presenting as Bladder Outlet Obstruction

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Abstract

Infants of diabetic mothers (IDM) are at increased risk of congenital malformations due to the teratogenic effect of maternal hyperglycaemia. Hydrometrocolpos, a developmental cystic dilatation of the uterus and vagina arising from the accumulation of cervical secretions and concomitant genital outflow tract obstruction is rare, occurring either in isolation or as part of recognised syndromes.

We report a case of hydrometrocolpos, an unreported condition in an IDM, presenting at birth as an intrabdominal mass manifesting as bladder outlet obstruction with secondary urinary tract infection to highlight its isolated occurrence in these high-risk infants that commonly present with systemic and other multiple congenital defects.

Keywords: Hydrometrocolpos, infant of diabetic mother, bladder outlet obstruction.

Introduction

Infants of diabetic mothers (IDMs) are at increased risk of morbidity and mortality including malformations, notably, lumbosacral agenesis and cardiac anomalies.[1,2] Congenital abnormalities of the genitourinary system, though not uncommon, mostly involve the renal system, with documented reports of genital anomalies observed in only three IDMs manifesting as aphallia in a male infant and anal atresia with rectovaginal fistula in two female neonates with other multiple congenital anomalies.[3] No reports of isolated genital anomalies in an IDM have been documented.[1,4]

Hydrometrocolpos, a developmental cystic dilatation of the uterus and vagina resulting from a combination of increased cervical fluid secretions and obstruction of the genital outflow tract is rare, with an incidence of 1 in 16,000 female births.[5] Vaginal atresia, vaginal septum, imperforate hymen [6,7] or urogenital sinus with urine collection linked with Bardet–Biedl syndrome, McKusick–Kaufman syndrome, Ellis–van Creveld syndrome, and Pallister–Hall syndrome [8] are common associations. The presentation may be asymptomatic, detected in adolescence [9] but may also be diagnosed prenatally as a pelvic mass with-the earliest reported case at 25 weeks' gestation,[10] or at birth

with abdominal distension and obstructive symptoms.[11–14]

We report a case of hydrometrocolpos, an unreported condition in an IDM, to highlight its isolated occurrence in these high-risk infants that commonly manifest with systemic and other multiple congenital defects.

Case Summary

A 3-day old female neonate (Figure 1) was referred to our facility with progressive abdominal distention and failure to pass urine since birth but had passed meconium on the 2nd day of life. The mother is a 22yr old para 3 + 0, in a non-consanguineous marriage. She was diagnosed diabetic 4 months before the conception of this baby and was managed with oral metformin with good glycaemic control. Metformin was stopped at 8 weeks of gestation when the pregnancy was confirmed. She was subsequently on dietary modification throughout her pregnancy with fair glycaemic control. Her antenatal visits were at a private facility and she was regular on her follow-up visits. A prenatal ultrasound scan at 30-weeks' gestation showed features of bladder outlet obstruction and foetal macrosomia. There was no history of maternal febrile illness, body rashes, exposure to radiation or ingestion of traditional/unprescribed medication. The mother does not smoke or ingest alcohol. Delivery was via elective caesarean section at a private hospital.

On examination, the baby weighed 4kg and had no dysmorphic features. She was afebrile (36.8ºC), not pale or cyanosed, anicteric, with a respiratory rate of 46 breaths per minute and oxygen saturation of 92%. The abdomen was grossly distended with an abdominal girth of 38cm, measured 8cm from the xiphisternum, visible veins draining upwards and no area of tenderness. There was a palpable, smooth, firm, non-tender intra-abdominal mass extending from the pelvis to the epigastrium measuring 14cm along its longitudinal axis. Other intra-abdominal organs including the kidneys were not demonstrable due to the swelling precluding palpation. Bowel sounds were audible posteriorly. The anal orifice was patent and normal in position. Examination of the genitalia revealed normal labia majora and minora, normal-sized clitoris, with a urethral opening 1cm below the clitoris and two slit openings on either side of the urethra representing the paraurethral glands (skene's gland). There was no vaginal orifice. All the limbs had normal muscle bulk, posture, tone, reflexes and power, with normal spine and digits. Other examination findings were normal.

A nasogastric tube was passed, which drained a yellowish effluent. Urethral catheterization was difficult and 20mls of turbid urine was drained with no significant change in the position and size of the mass. Urine microscopy revealed many yeast cells, 4-6 red blood cells, 1-2 pus cells and a few epithelial cells per high power field and culture yielded *Candida albicans.* A diagnosis of obstructive uropathy with urinary tract infection was made. An abdominal ultrasound scan showed a well-circumscribed intra-abdominal cystic mass originating from the pelvis and extending into the vaginal vault, with features of bilateral hydronephrosis. Figure 2 shows the abdominal ultrasound scan and X-ray findings. Magnetic Resonance Imaging of the abdomen showed gross enlargement of the uterus containing a uniform hyperintense fluid on T2 and hypo-intensity on T1 sequence. There was compression with effacement of adjacent myometrium with no focal mass seen. The urinary bladder was markedly compressed and anteriorly displaced while the rectum was intact. Both kidneys were normal in position with a grossly dilated pelvicalyceal system (Figure 3). Other investigations revealed no complications of IDM, with normal glycaemic control, electrolytes and blood counts. A definitive diagnosis of obstructive uropathy secondary to hydrometrocolpos with vaginal septum was made.

Expressed breast milk was given orally at 50% maintenance due to poor tolerance from pressure effect on the stomach with the balance fluid

requirement given as intravenous fluids. Antibiotics and fluconazole were also instituted.

Surgery was done on day 11 of life, under general anaesthesia via a suprapubic transverse incision. Operation findings include a slightly distended bladder despite catheterization, with a huge cystic mass posterior to and adherent to the urinary bladder wall. Needle aspiration of the cystic mass revealed a slightly turbid serous fluid, which was drained using a foleys catheter inserted via a transverse incision on the anterior wall of the mass. Approximately, 350mls of fluid was drained and the catheter was left in-situ. It remained active for one week, with the resolution of symptoms including normal urine flow and renal function test. A repeat pelvic scan at 3 months showed normal-sized kidneys with mild fluid collection in the dependent part of the uterus. The baby was referred to the gynaecologist for vaginal septum resection and vault dilatation scheduled at 6 months of age.

Discussion

The presentation of the new-born with abdominal distension and features of urinary obstruction detected prenatally, similar to reports by several authors,[11,13,14] coupled with the absent vaginal orifice, radiologic evidence of cystic uterine mass and normal lower urinary system confirmed the diagnosis of hydrometrocolpos with associated vaginal obstruction.[10] The clinical presentation of this condition in the neonatal period varies, depending on the size of the uterine fluid collection, and its most severe form is dominated by regional obstructive symptoms involving the urinary [13,14] and rarely the digestive system.[11] Although hydronephrosis was noted in the index case and it is common in IDMs, its occurrence, in this case, is unlikely to be due to the teratogenic effect of diabetes on the foetus[2] but rather secondary to obstruction in urinary flow from the pelvic mass as evidenced by its resolution following treatment on the subsequent scan at 3 months. Other secondary pressure effects include pulmonary hypoplasia, respiratory distress and leg swelling.[10] No other congenital anomalies were noted in our index case, in contrast to other reports.[3,8]

The association between maternal diabetes and congenital malformations is well established, [1,2,4] although, this increased risk in mothers who have a well-controlled disease is lower. While the mother narrated relatively good glycaemic control during the second trimester when compared to her glycaemic control prior to conception, foetal macrosomia, a common occurrence in 15-45% diabetic pregnancies[15] was noted in this neonate, which may suggest the glycaemic control may not have been adequate during pregnancy as reported historically by the mother. Furthermore, the teratogenic effect, type and severity of malformations due to prenatal exposure to maternal hyperglycaemia has been correlated with the period of organogenesis in the first trimester, [4] a period during which the mother was still on medication. Data on pregnant women treated with oral hypoglycaemic agents are not well documented.[2] There is no documented evidence of an association or increased risk to congenital malformations in the foetus following exposure to metformin in-utero.[16] Conversely, Soler *et al* in 1976 as reported by Mills[2] documented an 8.7% risk of malformation in infants of women receiving oral hypoglycaemics in the first trimester. However, the sample size was small for generalization. IDMs have a higher risk of congenital malformations than their counterparts born to non-diabetic mothers.[2] The difference in the relative risk for each system affectation suggests that various organs have varying susceptibility threshold to the teratogenic potential of maternal hyperglycaemia, with single organ affection which was seen in this infant observed in 70% of cases.[4]

The diagnosis of hydrometrocolpos is uncommon in the neonatal period due to its rarity, and it is usually detected in adolescence, presenting with cyclical lower abdominal pain and/or amenorrhoea.[14] Prenatally, differential diagnoses include pelvic cystic pathologies like adnexal cysts, anterior meningocele, pelvic component of sacrococcygeal teratoma, meconium pseudocyst,

dilated bowel, mesenteric cyst, rectal duplication, cystic neuroblastoma, or bladder duplication. [5,17] A high index of suspicion with emphasis on detailed physical examination and radiologic investigations are necessary to make a definitive diagnosis in the new-born. In our case, MRI was sufficient to make the diagnosis before surgical intervention, with complete resolution of symptoms post-operatively.

For isolated cases of hydrometrocolpos, the prognosis is good with surgical intervention,[10] as demonstrated in this case. Other factors such as associated malformations, background medical conditions, invasive surgical procedures can adversely affect the outcome.

Conclusion

Hydrometrocolpos in the neonatal period should be ruled out in any female new-born with a pelvic mass with or without obstructive symptoms. Its isolated occurrence in an IDM needs further evaluation for a definitive association, to aid in counselling parents on the risk of recurrence in future pregnancies, as no previous reports have been documented.

Declaration of patient consent

The authors certify that appropriate informed and written consent was obtained from the parents for clinical information to be reported for academic purposes in the journal.

Conflict of interest

Authors have no competing interests to declare.

Financial declaration

Nil

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