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Original Research Article

Prenatal ultrasound diagnosis of anterior abdominal wall defects in sub Saharan Africa; simple but often missed

Andrew H. Shitta[1]*, Mercy W. Isichei[1], Ezekiel D. Dung[1], Solomon D. Peter[1], Michael B. Ode[2], Isaac A. Jatau[3], Onyedikachi N. Ottah[1], Alexander F. Ale[1]

[1]Department of Surgery, [2]Department of Orthopaedics and Trauma, University of Jos, Jos University Teaching Hospital, Jos, Plateau State, Nigeria
[3]Department of Surgery, Federal Medical Centre, Keffi, Nasarawa State, Nigeria

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***Correspondence:**
Dr. Andrew H. Shitta,
E-mail: shittosis@yahoo.co.uk

ABSTRACT

Background: Congenital anterior abdominal wall defects (AAWD) is a spectrum of abdominal wall defects that includes omphalocele, gastroschisis, bladder exstrophy, cloacal exstrophy, prune belly syndrome and pentalogy of Cantrell. Early Prenatal diagnosis of AAWD provides opportunity for abnormal karyotypes screening and planned delivery in a specialized centre. Ultrasound can detect these defects during pregnancy. This study aims to evaluate the detection rate of AAWD during routine obstetric ultrasonography in our region.
Methods: A retrospective study of all patients that presented with AAWD to our centre from January 2008 to July 2020. Data included patient's age, sex, birth weight, diagnosis, resuscitation time, outcome, maternal age, parity and antenatal ultrasound scan (USS) records. Antenatal USS before 12 weeks only, were excluded. Data analysed using excel.
Results: Of the 140 with AAWD, 84.29% had omphalocele, 10% gastroschisis, 2.14% prune belly syndrome and 0.71% each with bladder exstrophy, cloacal exstrophy and pentalogy of Cantrell. There were 123 booked pregnancies. Majority (112) had antenatal care elsewhere while 11 attended our Centre. Ultrasonography of 108 pregnancies scanned at12 weeks or beyond, had 4 confirmed prenatal diagnosis of AAWD. All done in our centre. Mean gestational age at diagnosis was 24weeks. Outcome was rupture1 (25%) and 25% mortality (prenatally diagnosed) and 51.92% mortality for patients with missed diagnosis.
Conclusions: Our obstetric ultrasound detection rate of AAWD is very low. There is a need for improvement in training to improve perinatal care of these defects.

Keywords: Prenatal diagnosis, Ultrasonography, Anterior abdominal wall defects

INTRODUCTION

Congenital anterior abdominal wall defect is a spectrum of abdominal wall defects that comprises of omphalocele, gastroschisis, bladder exstrophy, cloacal exstrophy, prune belly syndrome and pentalogy of Cantrell The commonly encountered defects are omphalocele and gastroschisis with an incidence of 1.13:10,000 and 2.65:10,000 Live births respectively.[1] These two common anterior abdominal wall defects can be detected on ultrasound within the first trimester of pregnancy.[2]

Early Prenatal diagnosis of anterior abdominal wall defect provides opportunity for carrying out prenatal screening and detection of associated anomalies or chromosomal aberrations so that parents can be offered early

counselling; preparing the parents for the possibility of the birth of a handicapped child and allowing for a planned delivery in a specialized center for the care of such a newborn.[1-3] These benefits of early diagnosis offer a better outcome in the management of anterior abdominal wall defect.

Ultrasonography, a readily available tool is cheap and has a high ability to detect anterior abdominal wall defects during early pregnancy.[2] Prenatal ultrasonography has been reported to have up to 75%-80% sensitivity and 95% specificity in diagnosing theses defects.[3,4] In prenatal ultrasonography, omphalocele can be seen as herniation of visceral content through a midline defect covered by a thin membrane with apical insertion of the cord.[5] Most commonly is ascites seen within the sac Gastroschisis appears as normally sited cord with visualization of a small defect commonly to the right of the cord with herniation of free floating intestines without a membrane.[6,7] Pentalogy of Cantrell can be diagnosed as early as the 10th gestational week, and is visualized as a supraumbilical defect with an ectopia cordis.[8,9] Bladder exstrophy can be detected on ultrasonography as a solid mass between the insertion of the cord and the fetal genitalia with failure to visualize the bladder in the presence of normal kidneys and collecting ducts.[10,11] Prune belly syndrome is commonly diagnosed prenatally by the 2nd and 3rd trimester.[12] Pathognomonic sonographic feature is an elongated tortuous dilated ureters.[5]

Perinatal ultrasonography for the diagnosis of anterior abdominal wall defects has its own challenges when carried out on a complicated pregnancy that interferes with ability to visualize the structures. The presence of oligohydramnios for instance, could pose a challenge in clearly identifying the anterior abdominal wall defects even in the hands of an experienced sonographer. Other imaging techniques with better resolution is needed to clinch the diagnosis. MRI has been found to offer better resolution in the presence of oligohydramnios.[13]

Other non-radiologic investigation that can help in increasing the strength of diagnosis in suspected case of anterior abdominal wall defect is elevated maternal serum alpha fetoprotein (MSAFP). High levels of MSAFP in a pregnant woman is an indication for ultrasonic evaluation of the fetal ventral wall.[5] Although MSAFP level could serve as a first line indication for further examination, in our setting, standard laboratory that can carry out this test is not readily available. Ultrasound on the other hand has become available and is fast becoming a standard of care in the prenatal stage of pregnancy. There are portable ultrasound machines in use all over Africa. Ultrasound machine is easy to use and can be mastered quickly when frequently used. This tool can assist in early diagnosis of anterior abdominal wall defect in our setting. Compared to MRI that is expensive, not portable and requires a high technical skill, ultrasonography is a much better option.

Despite the significant contribution ultrasound plays in prenatal diagnosis of anterior abdominal defect, which improves management and consequently, better prognosis for patients thereby reducing perinatal mortality, it is not adequately utilized. It is worthy to note that ultrasonography is routinely required during prenatal care as part of efforts to increase early diagnosis of fetal abnormalities and reducing perinatal mortality through early intervention. But, records show that this simple, cheap and readily available tool it is not fully utilized in this region that contributes more than half of global perinatal mortality.[14] In the absence of prenatal diagnosis, management outcome for infants born with this defect is attended with a very high mortality rates especially in moderate and major defects.[15]

This study aims to evaluate the detection rate of anterior abdominal wall defects during routine obstetric ultrasonography in our region.

METHODS

This is a retrospective study of data collected from all patients that presented with anterior abdominal wall defects to the paediatric surgical unit at the Jos university teaching hospital from January 2008 to July 2020. The paediatric surgical unit has 4 consultants serving a paediatric population estimated to be 1.7 million within the state and parts of Bauchi, Kaduna, Nassarawa and Benue states with estimated population of under 20 years to be 8.4 million.[16]

Information collected included patient's age, sex, birth weight, type of defect and associated malformation/anomaly, time to presentation and resuscitation and outcomes (rupture, sepsis, discharged or mortality). Maternal data included maternal age and parity, pregnancy and delivery history included booking date, place of booking, gestational age at booking, antenatal ultrasound scan (USS) and gestational age at sonography, prenatal diagnosis from USS report. We considered new born delivered at our centre as in-born while those referred to our centre as out born. All diagnosis were confirmed by the consultants

Those included, were those who had antenatal USS at least once at ≥12 weeks GA. Any index pregnancy that had USS scan at less than 12weeks GA only, was exclude.

Data was inputted and analysed using excel worksheets.

RESULTS

There were 140 new born babies who presented to our facility with the diagnosis of anterior abdominal wall defects within the study period with a male to female ratio of 1:1.1. Of this number 118 (84.29%) had omphalocele, 14 (10%) gastroschisis, 3 (2.14%) prune belly syndrome (Figure 1) and 1 (0.71%) each with bladder exstrophy, cloacal exstrophy and pentalogy of Cantrell.

Figure 1: A newborn with prune belly syndrome.

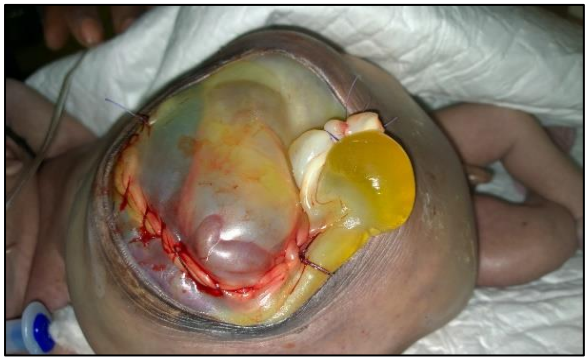

Figure 2: A prenatally diagnosed macrosomic newborn with omphalocele that had inadvertent laceration of the sac during caesarian sectioning.

There were 123 booked pregnancies at mean gestational age of 17.8 weeks while 17 cases were product of un-booked pregnancies. Health care facilities visited were 18 (14.63) booked in a primary health care, 58 (47.15) secondary health centre, 36 (29.27%) private health facilities while 11 (8.94%) had antenatal care in our Centre. History of obstetric ultrasonography showed 8 (6.5%) had a single scan at gestational age less than 12 weeks, 108 (87.81%) had at least a scan/ repeat at gestational age of 12 weeks or beyond and 7 (5.7%) of those who booked never had a scan. Of the 112 who booked outside of our centre, 72 had obstetric ultrasonography in private facilities and 32 in secondary health facilities. Only 4 (3.7%) patients had a prenatal diagnosis of anterior abdominal wall defects. All were diagnosed as having omphalocele but one turned out to be gastroschisis. All diagnosis was done at our centre (detection rate of 36.36%). Mean gestational age at diagnosis was 24 weeks (range 17 weeks to 31 weeks). Three deliveries were via spontaneous vaginal delivery (SVD) and 1 through an elective Caesarian Section on account of macrosomia (syndromic Beckwith Weidman Figure 2). This patient died soon after birth. There was accidental laceration of the sac during caesarian sectioning). We had a rear case of pentalogy of Cantrell with a prenatal ultrasound scan at 10 and 16 weeks done at a secondary health care facility that suggested a twin gestation with 1 non-viable gestational sac. There was a failed attempt to abort this non-viable gestational sac at the first scan. Pregnancy was followed up with repeated scans and was delivered at term via an elective CS. We also lost this baby post operatively. There were 11 Inborn and 129 out born deliveries. Time of presentation and resuscitation for the out born deliveries was 34.6 hours and 0.5 hour for the inborn.

Of the 104 (96.3%) who were missed prenatally, 42 (40.38%) had rupture of their omphalocele, 37 (35.38%) sepsis with 54 (51.92%) mortalities. Morbidity among the prenatally diagnosed included rupture/laceration and sepsis (1, 25% each). Mortality rates for the inborn and out born were 18.2% and 51.92% respectively.

DISCUSSION

Over 2/3rd of the pregnancies were booked and had access to obstetric ultrasound. This compares with the data from national demographic survey of 67 %, even though our value was much higher, likely because majority of our patients had access to secondary and private health facilities where ultrasound machine is readily available.[17]

Majority of cases in our study were missed during obstetric sonography. All prenatally diagnosed cases were done in our teaching hospital. A study done in Cameroun also found a low rate of prenatal diagnosis of visible congenital malformations of just 21%. This was attributed to poor training.[18] Similarly, a study here in Nigeria (Ilorin) showed a prenatal ultrasound diagnostic Rate of1.8% in fetuses with gastroschisis and omphalocele despite high rate of obstetric scans (91.1%).[19] This quantum of missed diagnosis could be explained by lack of trained and experienced personnel in identifying abnormal obstetric scan. The emphasis on; detecting fetal viability, estimation of gestational age, sex determination and expected date of delivery, placental localization and multiple gestation has also not helped in prenatal diagnosis of these cases even if they exist.[14] This can be seen in the case of the fetus with pentalogy of Cantrell as the omphalocele sac was diagnosed as a second nonviable gestational sac with resultant missed of an overt sonographic features of this rare anomaly in an inexperienced hand. However, in more developed climes, prenatally diagnosed anterior abdominal wall defect detection rate and accuracy can be as close to 100%, based on the availability of scanning equipment and experience of the sonographer.[20]

Our study also showed higher morbidity and mortality rates among the missed diagnosis and out born compared to the prenatally diagnosed and inborn. Nasir et al, in a study of 395 prenatally diagnosed gastroschisis found significant rate of morbidity in infant born outside a perinatal centre and concluded that delivery location is a significant independent predictor of complications in infants with gastroschisis.[21] Studies in Zimbabwe also showed that deliveries at home and away from paediatric specialist centres increased the risk of complications in patients with gastroschisis.[22]

All the patients who were out born did not have a prenatal diagnosis. There was a higher rate of sepsis, membrane rupture among them and mortality as well, compared to the inborn and prenatally diagnosed cases. This may be explained as the undiagnosed pregnancy was not managed as high risk and therefor, there were no special measures taken to ensure safe delivery. Deliveries of such babies were never prepared and occurred either at home, or in health facilities with no experience and far from a specialized paediatric centre as ours. Also, mean time to presentation and resuscitation of the out born was more than 30 times longer than the inborn and prenatally diagnosed. These babies were most likely to be poorly resuscitated before transfer/referral. Stevens et al found out that resuscitation is a significant factor in the outcome.[23]

Limitations

There was no data on the level of training of the sonographers outside our facility.

CONCLUSION

Our obstetric ultrasonic detection rate of fetal anterior abdominal wall defects is very low as in other sub Saharan regions. There is a need for improvement in training so as to improve on this detection rate. This will go a long way in contributing to improving the perinatal care and thereby reducing perinatal morbidity and mortality of these patients in our region.

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