A Review of Five Cases of Fetal Congenital Anomalies Diagnosed during Prenatal Ultrasound: Management Problems and Limitations

ABSTRACT

Congenital/fetal anomalies are structural, functional, and/or biochemical–molecular defects present at birth whether detected at that time or not. Prevalence of congenital fetal anomalies in developing countries especially in Africa is underestimated. They are seen in 2 to 3% of newborns and about 94% of severe birth defects are seen in low-resource countries. Currently, they account for 20 to 30% of perinatal mortality in developed countries. Their contribution to perinatal mortality may be much higher in developing countries. They are also an important cause of maternal mortality in developing countries because they cause prolonged obstructed labor, ruptured uterus, postpartum hemorrhage, and sepsis. Ultrasound is a very important tool in prenatal diagnosis, but ultrasound usage faces a lot of challenges in Africa. When congenital anomalies are diagnosed in Africa, management becomes a challenge as patients may not even accept the diagnosis or the line of management and this leads to catastrophic complications for the mother and her unborn infant. To overcome these challenges, various measures must be taken, such as training for practitioners, provision of equipments, community enlightenments concerning causes, possible management, and prevention.

This review is meant to highlight some of the challenges facing prenatal diagnosis and management of prenatally diagnosed congenital fetal anomalies in low-resource environments and suggest the way forward.

Keywords: Anomalies, Congenital, Prenatal, Review, Ultrasound.

INTRODUCTION

Congenital/fetal anomalies are structural, functional, and/or biochemical–molecular defects present at birth whether detected at that time or not.1 Prevalence in developing countries especially in Africa is underestimated because of deficiencies in diagnostic capabilities, lack of reliable medical records, and statistics.2 Congenital anomalies are seen in 2 to 3% of newborns.3 About 94% of severe birth defects are seen in low-resource countries, particularly in Africa.2 Major fetal congenital anomalies are malformations that affect fetal viability and/or quality of life, and minor anomalies are malformations that are definitely present, but are minimal and usually have no functional significance.3 About 20% of anomalies in live-born infants are based on defective gene, 10% are due to chromosomal abnormalities, and 10% are due to exogenous injury to the conceptus.6 Some 60% of all congenital anomalies are of indeterminate cause or are multifactorial in origin (due to hereditary or environmental factors).6 In Africa, all these factors may play a role, for example, nutritional deficiencies (folic acid, iodine), maternal diseases (diabetes, thyroid diseases), maternal exposure to harmful substances in pregnancy (lead, dichlorodiphenyltrichloroethane, radiations), maternal infections in pregnancy (rubella, cytomegalovirus, Zika virus, syphilis), use of herbal concoctions as medication which may potentially contain teratogenic agents in pregnancy, and pregnancy at or above 35 years.7 Other factors include poverty, and low level of health care available to communities.8,9

The significance of congenital fetal anomalies lies in the fact that they are a major cause of perinatal mortality. In Africa, they also expose women to unnecessary risks, such as obstructed labor, puerperal sepsis, postpartum hemorrhage, and also adds to the high burden of maternal mortality. Because of improvement in other areas of perinatal care, birth defects are the single most common cause of perinatal mortality in developed countries, accounting for 20 to 25% of perinatal deaths.6 Effective prenatal care involves screening examinations in pregnancy among other measures. Among the various tests now offered to pregnant women, ultrasound has the broadest diagnostic spectrum.6 There is no modality that can detect as many abnormalities during pregnancy as ultrasound.6 A priority goal in screening is early detection of major fetal anomalies.5 Early detection will provide an opportunity for early counseling and intervention in the form of treatment or pregnancy termination where available and legally possible.
In Africa, we have a complex situation as it concerns issues of prenatal diagnosis and management of congenital fetal anomalies as the situation is confounded by illiteracy, poverty, and unavailability of trained personnel and equipment for proper diagnosis and management. Prenatal ultrasound diagnosis of congenital defects is one of the black points of African ultrasonography.

The number of prenatally diagnosed congenital anomalies is very low. Malformation detection rate does not exceed 20%. This is often too late in pregnancy to enable meaningful intervention to impact on positive outcome. Most times diagnosis is in labor or even at cesarean section or laparotomy when complications have occurred. Thus, prenatal diagnosis of congenital fetal anomalies is opportunistic and the largest proportion of pregnant women are excluded from the opportunity.

This review is meant to highlight some of the challenges facing prenatal diagnosis and management of prenatally diagnosed congenital fetal anomalies in low-resource environments and suggest the way forward.

**CASE REPORTS 1**

A 37-year-old unbooked, G_{7}P_{6}+0 with six living children, with her last menstrual period on January 27, 2015, presented with complaints of reduced fetal movements. She had no antenatal care in all her previous pregnancies. She had no significant complaints in her current pregnancy and has not been on any medication except for herbal concoctions which she started taking since she found that she was pregnant. She had no history of liquor drainage or bleeding per vaginum. On examination, she was not pale, was not febrile, and was not jaundiced. Her vital signs were within normal limits. Symphysiofundal height was 24 cm, fetal heart sound was present, and there were no palpable uterine contractions.

An obstetrics ultrasound (done with a two-dimensional ultrasound) revealed a viable fetus, biparietal diameter = 6.53 cm equivalent to 26 weeks + 4 days, expected date of delivery (EDD) = October 26, 2015, with gross polyhydramnios (single vertical pool = 13.7 cm), gross fetal ascites, pleural effusion, multiple cystic masses on the posterior aspect of the fetal neck, and congenital hydrocele. She was counseled on the findings and options of management. She said she needed to discuss with her husband before taking a decision. She went home and came back after 10 days. A day before her second presentation she started draining liquor and just before presentation she started bleeding per vaginum. A diagnosis of placental abruption was made and she was resuscitated and 3 hours later delivered of a fresh stillborn male infant with features of cystic hygroma. She later developed severe postpartum hemorrhage and was treated with 5 pints of blood and intravenous fluid.

**CASE REPORTS 2**

A 20-year-unbooked G_{1}P_{0}+0, unsure of her last menstrual period but said to be about 28 weeks pregnant, came for routine antenatal booking and had no complaints. On examination, she was not pale, afibrile, not jaundiced. Her vital signs were stable. Symphysiofundal height was 26 cm and fetal heart sound was present. An obstetrics scan revealed a viable fetus, at 29 weeks 6 days, EDD = January 16, 2016, with a cranial defect containing fluid. A diagnosis of encephalocele was made. She was counseled on the findings and the need for evaluation at intervals. She was booked and pregnancy was monitored till 37 completed weeks. She went into spontaneous labor and delivered a live female baby with a cystic swelling on the occipital region. A diagnosis of occipital encephalocele was made with no other apparent anomaly. Baby was handed over to the neonatologist.

**CASE REPORTS 3**

A 47-year-old unbooked G_{10}P_{8}+1 with eight living children, unsure of her last menstrual period but was said to be 8 months pregnant, had history of fever when she was 2 months pregnant. She was admitted for 14 days and treated for the fever at a local hospital. She presented with history of liquor drainage and absent fetal movements. On examination, she was not pale but febrile to touch (38.5°C). Her pulse rate was 108/min, blood pressure was 110/80 mm Hg. Symphysiofundal height was 32 cm, the fetal lie was transverse, there was no fetal heart tone and no uterine contractions. Sterile speculum examination revealed a closed cervix and no evidence of liquor drainage. An obstetrics scan revealed a nonviable fetus in transverse lie at 30 weeks + 6 days of gestation, the fetus had a defective anterior abdominal wall with bowels and liver protruding through the defect and floating in liquor. A diagnosis of intrauterine fetal death and gastroschisis was made. The patient was counseled and she consented to pregnancy termination via cesarean section. A fresh stillbirth fetus was delivered with bowel and liver protruding through anterior abdominal wall defect with no other obvious anomaly.

**CASE REPORTS 4**

A 40-year-old unbooked G_{14}P_{10}+3 with eight living children had two infants with congenital anomalies in her previous pregnancies. She had been taking medications to prevent a recurrence of congenital anomalies which she bought at a chemist. She presented at 29 weeks of
gestation with excessive fetal growth and respiratory difficulty. On examination, she was not pale and not jaundiced. Symphysiofundal height was 44 cm, there was positive fluid thrills and absent fetal heart tone. An obstetrics scan revealed a single nonviable fetus with anencephaly, polyhydramnios with no other detectable anomaly. A diagnosis of anencephaly and intrauterine fetal death was made. Patient was counseled for pregnancy termination but refused to accept the diagnosis. She went to another hospital where they ruptured the membranes and commenced her on oxytocin infusion. She was referred back to our hospital when she started bleeding per vaginum and the fetal hand had prolapsed per vaginum. A diagnosis of hand prolapsed and ruptured uterus was made. She was resuscitated and had exploratory laparotomy. The uterus was repaired and bilateral tubal ligation was carried out. A female fetus with anencephaly weighing 2.0 kg was extracted Figure 4.
Fig. 3: Case 3

Fig. 4: Case 4
CASE REPORTS 5

A 28-year-old booked primigravida presented for routine obstetrics scan. The scan revealed a live female fetus at 31 weeks + 5 days with a unilateral ovarian cyst with no other obvious anomaly. She was counseled on the findings and was advised for serial repeat scan to monitor the cyst. She had four follow-up scans which all revealed a progressive reduction in the cyst diameter. The last scan was done at 37 weeks and no cyst was seen on the scan. Patient went into spontaneous labor and delivered a live female infant with good Apgar score with no obvious anomaly. A scan a week after delivery did not reveal any ovarian cyst Figure 5.

DISCUSSION

Prenatal diagnosis of congenital anomalies is still at infancy in most developing countries as it is limited by lack of trained and experienced practitioners and absence of proper equipments. It is also affected by lack of universal acceptability and availability due to poverty, illiteracy, and deeply entrenched cultural norms, which are inimical to application of modern technology in obstetrics care. The cases presented here vividly show that most women carrying fetuses with congenital anomaly do not book for antenatal care which would have provided them with the opportunity for early prenatal diagnosis and appropriate management including termination of pregnancy where it is allowed by law. All the patients discussed presented late in pregnancy and came to seek for care because they had one problem or the other and would have stayed to deliver at home if they had no complications, only to discover that the infants have anomalies. Only the last patient was booked for antenatal care and presented for routine obstetric ultrasound without any complaints. She also complied with instructions for follow-up until delivery. All the cases presented had major anomalies except the last one which had a minor anomaly, with the pregnancy ending with good outcome. It can be concluded that ultrasound application for fetal surveillance is one of the most important tools for evaluation of fetal well-being and diagnosis of different, major and minor, anomalies which if detected on time can rescue the mother and fetal life by changing the way of managing the pregnancy and delivery. This is seen in the last case where a minor anomaly was detected but on meticulous follow-up repeat scan the anomaly disappeared. Complications were observed in the first and fourth cases. In the first case, the patient refused to come back until 10 days when she developed placental abruption which put her life at risk and even when she delivered she further developed postpartum hemorrhage. This showed that it is not enough to make the right diagnosis, it is equally important to convince the patient for timely and correct intervention before the onset of possible complications. In the fourth case, again the patient refused to accept the diagnosis and the option of management given to her. She went to a lower level hospital where she was given an inappropriate treatment and as a result it led to uterine rupture. These demonstrate how patient behavior affect management outcome.

In low-resource environments, it is not enough to make diagnosis of congenital anomalies. Diagnosis must be complemented with proper treatment which will reduce maternal and prenatal complications. However, this is not always possible in low-resource environment as patients may sometimes refuse to accept the diagnosis or refuse to accept the line of management. The solution to this is to create community awareness about the causes and available management options and methods of prevention. The prerequisite to achieving these is providing facilities where prenatal diagnosis can be made, training practitioners who will be manning these facilities, providing appropriate equipment, and
providing counseling and other support services. The practice must also be regulated which will ensure safety of patients by observance of ethical principles.

**CONCLUSION**

Congenital anomalies are known to significantly contribute to perinatal mortality and indirectly to maternal mortality as well, hence the need to entrench prenatal diagnosis in our health care delivery system. However, prenatal diagnosis and management of congenital anomalies face a lot of challenges in low-resource countries. These challenges include lack of properly trained practitioners, lack of proper equipment and facilities. On the other hand, community awareness is poor as regards the causes of congenital anomalies, methods of management, and prevention. The consequence is that congenital anomalies continue to cause perinatal morbidity and mortality and sometimes maternal morbidity and mortality. Creating awareness and training practitioners will go a long way in addressing these challenges.

**REFERENCES**