Polyhydramnios Associated with Multiple Congenital Malformations: Case Report

O.A.O. Oloyede, M. A. Lamina, O. O. Adetoro

SUMMARY

A case of polyhydramnios associated with multiple congenital abnormalities in the fetus of a 25 year old primigravida, at 33 weeks gestation is reported. She presented with complaint of excessive abdominal distension and ultrasound scan revealed severe polyhydramnios (AFI 46.1cm) with multiple congenital malformations. The patient was delivered of a fresh stillbirth with multiple congenital anomalies.

Niger Med J. Vol. 49, No. 1, Jan. – Mar., 2008: 24 – 26.

***Keywords*: Polyhydramnios, Multiple congenital malformations (MCA), Amniotic fluid index (AFI)**

INTRODUCTION

A normal volume of amniotic fluid is an indicator of good foetal health[1]. It rises linearly from early gestation till about 32 weeks, when it remains constant till term and thereafter declines[1]. Polyhydramnios is defined as increased or excessive amniotic fluid volume, or an amniotic fluid index greater than three standard deviation or the 97.5 percentile for gestation[1, 2]. It is categorized into 3 categories, with the mild form being about 80%, moderate about 17% and severe about 5%[1]. In the second trimester and beyond, when the condition has more clinical significance, the incidence is about 1 in 200 pregnancies[1]. Its association with congenital malformations are well documented. As whole, congenital foetal abnormalities contribute about 20% of causes of polyhydramnios, while in severe polyhramnios, approximately 75% are caused by foetal abnormalities[1]. We report this case of severe polyhydramnios associated with multiple congenital abnormalities first because it is rare in our environment and second to draw emphasis on the importance of a careful foetal survey in cases of severe polyhydramnios and referral to appropriate centres for better management.

CASE REPORT

Mrs A.A a 25-year old pregnant Nigerian, primary school teacher was referred to our centre at 33 week gestation with a working diagnosis of polyhydramnios. Pregnancy was primarily booked at a maternity home where no antenatal investigations had been conducted. She has not received tetanus toxoid.

From: Dept of Obstetrics and Gynaecology, Obafemi Awolowo College of Health Sciences, Olabisi Onabanjo University, P.M.B 2001, Sagamu

Correspondence: Oloyede O. A. O, E Mail: oloyedeoao@yahoo.com

Gynaecological history revealed 5 years primary infertility, which was managed with traditional herbal drugs, till she achieved present pregnancy. Pregnancy was however confirmed at 12 weeks, while she was still on the herbal medications drugs. Also, she was treated for forunculosis at about 12 weeks of pregnancy, with herbal concoctions. She is the 7th of 9 children and neither smoked cigarette nor drank alcohol. There was no family history of congenital malformation or diabetes mellitus. She is not a known diabetic or hypertensive patient.

Physical examination in the antenatal clinic revealed a healthy looking pregnant woman, who was afebrile, not pale, anicteric and had no pedal oedema. Body temperature was 36.0^{0}C. Her blood pressure was 120/80 mmHg in sitting position and the pulse rate was 86 beats per minute, regular and of full volume. Abdominal examination revealed a grossly distended and shinny abdomen with a symphisioa fundal height of 40cm. The foetal lie and presentation could not be determined and the foetal heart was faintly heard at 138 beats per minute.

She was admitted for further evaluation and the results of the investigations are as below:

Packed cell volume (PCV): 29 %

Blood group: O Rhesus Positive

Urinalysis: Normal

Fasting Blood Sugar (FBS): 78 mg/dl

Foetal Ultrasonography

Brain / Central Nervous System

The BPD corresponds to a term pregnancy (93.3mm). There were features of incomplete cleavage of the forebrain, namely partial segmentation of the ventricles and cerebral hemispheres posteriorly. The thalami are incompletely fused (semilobar holoprosencephaly). The lateral ventricles are also dilated (RT 33.6mm: LT 30.4mm). This is consistent with severe hydrocephalus. In addition to these, there is a significant nuchal fold oedema. The spinal cord terminates at the level of the upper thorax, with appearance suggestive of degeneration below the level and gibbus?.

Heart

The 4 and 5 chambers view of a normal size heart visualized. Heart rate is 158 beats per minute.

Abdomen

Reduced gastric size (8mm x 9mm x 16mm). This may suggest a partial intestinal obstruction. ?Oesophageal atresia.

Musculoskeletal System

There is shortening of the extremities involving the entire limbs. The measurable FL of the left limb corresponds to 13 weeks pregnancy. [FL=16mm (range=59-69mm)]. No rib fracture. This form of osteoachrondrodysplasia is referred to as achondrogenesis type I, a variant of micromelia [severe limb shortening, short trunk, short chest and large head]. It is a lethal dysphasia with a birth prevalence of 1 in 40,000.

Amniotic Fluid

The Amniotic fluid index is 46.1cm. (Rt/Upper: 14.1cm, Rt/Lower: 8.9cm, Lt/Upper: 13.7cm, Lt/Lower:8.9cm).This confirms polyhydramnios.

Summary

1. Multiple Congenital Abnormalities (MCA)
2. Severe Polyhydramnios

The results of the investigations especially the ultrasound scan was discussed with her as well as the possible management options. She agreed to have the pregnancy delivered immediately.

She remained on admission, while plans were made for cervical ripening with misoprostol and induction of labour with pitocin. She was delivered after 72 hours of admission, of a fresh still female foetus with the following features:

Birth weight - 1.5kg
Length of baby – 27mm
Head circumference – 33mm (2.8-3.4)
Chest circumference – 24mm (21-27)
Abdominal girth – 29mm (2.6 – 3.4)
Large dimple on the back
Bilaterally absent humerus but short radius and ulna
Bilaterally short femur, tibia and fibula
Weight of placenta 0.3kg
4 litres of greenish coloured liquor (1000mls)

The parents were counselled to have post mortem autopsy carried out on the foetus, which was refused for undisclosed personal reasons. Spiritual counselling was done by the local priest and this improved her wellbeing in the postpartum period.

DISCUSSIONS

This case of polyhydramnios associated with multiple congenital malformations will be the first to be reported from this hospital, which is the referral centre for most health institutions within the state and environ cities. Although there are no local data on the prevalence and aetiology of polyhydramnios, it is however known that most cases are mild and idiopathic in origin[1, 2]. The moderate and severe forms as in this case can result from either a maternal or foetal disorder. Approximately 75% of cases of severe polyhydramnios are caused by foetal abnormality, while the rest may be of maternal origin or idiopathic[1]. The hallmark of diagnosis in this case was the combination of the clinical symptom of excessive abdominal distension and ultrasound evaluation of the amniotic fluid volume. In Mrs A.A, the AFI was 46.1cm, which confirms severe polyhydramnios. This method is more objective than the visual estimation of the amniotic fluid that may also be used.

In this report, multiple congenital malformations were detected on the ultrasound scan survey. Of particular aetiological significance is the suggestion of a partial intestinal obstruction, which has been reported as the most common type of foetal malformation associated with polyhydramnios and could also have been contributory in this case[1]. Although the precise type of native drugs ingested could not be ascertained, but the period of ingestion is when it could produce teratogenic effect on the developing foetus[3, 4, 5]. Consequently, the ingestion of native concoctions was a strongly considered aetiology in this case.

Another important finding in the foetus is the nuchal fold oedema, which is the second trimester equivalent of nuchal translucency. In the absence of other causes such as cardiac abnormalities, infection or genetic syndromes, its presence becomes a strong second trimester marker for chromosomal abnormality[6]. This possibility is further supported by the presence of multiple malformations. Down's syndrome is the commonest chromosomal abnormality associated with multiple congenital abnormalities[7]. Postnatal diagnosis of abnormal karyotype is advised in this case, but could not be done because of patient's refusal to accept the procedure. Similarly, she declined post mortem examination of the foetus. Socio- cultural and religious reasons are often times the reason for refusal, as congenital malformations are attributed to witch crafts and sorceries. This however is ideal and recommended to enable a definitive diagnosis of the aetiology to be made.

The management option considered in the case is pregnancy termination, in view of the multiplicity and severity of the malformations. The chance of extra uterine survival was considered reduced, while survivors would be affected by severe handicap that would affect quality of life. Hence, Mrs AA had induction delivery and was delivered of a fresh still foetus, which probably died during labour induction. In centres, where there are possibilities of corrective post natal surgeries, this option might be considered in the counselling and pregnancy continued till term for an improved management outcome. Post natal counselling is an important aspect of management that was offered to her, with emphasis on early pregnancy booking in the next pregnancy. This is because there is a chance of recurrence of congenital abnormalities in succeeding pregnancies depending on the cause and type of malformations involved. She was also advised to avoid ingestion of unprescribed medications and native concoctions of undetermined composition in subsequent pregnancy.

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