

Case Report

Importance of Prenatal Diagnosis in the Effective Management of the Hydrocephalic Fetus: A Case Report in the Douala General Hospital, Cameroon

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Abstract Congenital hydrocephalus is a well-known entity which is usually associated with other intracranial and extracranial malformations. The complications related to this condition range from intracranial (hypoplasia of the corpus callosum, encephalocele and arachnoid cysts) in 37% cases to extracranial malformations (myelomeningocele, renal agenesis, Fallot's tetralogy, septal defects, cleft palate, Meckel-Gruber Syndrome etc.) in 63% cases. The cornerstone to management is good prenatal follow-up with targeted neurosonograms and search for associated problems. During this time the decision for continuing or terminating the pregnancy is taken after patient counseling. Towards term, decisions will be made on the mode of delivery (vaginal or cesarean delivery) depending on the presentation of the fetus. Furthermore, it is important to avoid maneuvers that would lead to maternal and fetal jeopardy. Finally, we advise early referral of patients who are clinically suspected to carry fetuses with hydrocephalus to better equipped centers where sonographic confirmation and search for associated malformations could be done and appropriate and timely management of cases thereby reducing maternal-fetal morbidity and mortality.

Keywords congenital hydrocephalus; prenatal ultrasound; pregnancy; cesarean delivery

1 Introduction

Congenital hydrocephalus is etiologically a condition resulting from an abnormal increase in cerebrospinal fluid (CSF) within the ventricular and subarachnoid spaces of the brain, often resulting in enlargement of the cranial vault. The incidence of hydrocephalus has been reported to be 0.5 to 2 per 1000 total births [10]. In fetuses with non communicating hydrocephalus obstruction may occur in the

ventricular system in 43% of cases. Furthermore, fetuses with communicating hydrocephalus have obstruction beyond the ventricles; 38% in the subarachnoid space and 19% in the arachnoid granulations – Dandy Walker syndrome. There is a family history of hydrocephalus in 3.3% cases. Malformations associated with hydrocephalus could be intracranial or extracranial. There is a 37% association of hydrocephalus with other intracranial malformations especially hypoplasia of the corpus callosum, encephalocele and arachnoid cysts. Extracranial malformations occur in about 63% of cases ranging from myelomeningocele, renal anomalies (renal agenesis), cardiac anomalies (Fallot's tetralogy), gastrointestinal anomalies (agenesis of the colon) etc. [1]. In cases where an obstruction is not visualized, ventriculomegaly is the more appropriate term. Ventriculomegaly is "isolated" when the fetus has no other anomalies, except those that are a direct result of the ventricular enlargement. When the ventricles measure greater than 15 mm, many of those cases that appear isolated prenatally are ultimately found to have other abnormalities (e.g., Chiari malformations, neural tube defects, Dandy Walker malformations, agenesis of the corpus callosum, genetic syndromes) [13].

Initially, the ultrasound diagnosis of hydrocephalus was based on a biparietal diameter (BPD) measurement greater than 11 cm or a BPD 2 times greater than the thoracic diameter. However, the ultrasound diagnosis of fetal hydrocephalus has greatly improved with the advent of high resolution transducers including three dimensional (3D) images which permit accurate visualization and measurement of the ventricles [13]. Furthermore, since ventricular enlargement antedates head enlargement earlier diagnosis is possible especially in tertiary care centres

practicing perinatal medicine. Nowadays, hydrocephalus can be diagnosed very early in pregnancy on the basis of widening lateral ventricles with fetal biparietal diameter and head circumference still normal or without macrocrania [10]. In this paper, we report a case of undiagnosed fetal hydrocephalus that resulted in decapitation of the fetal head at vaginal breech delivery thereby causing fetal mortality and maternal morbidity. We also emphasize on the importance of prenatal diagnosis especially in the developing countries to look for associated malformations and need for early referral to tertiary care centres with the aim at reducing both maternal and fetal morbidity and mortality.

2 Case report

The patient is a 24 year old gravida 2 para 2 with one liveborn female baby and married to a nonconsanguineous 36 year old husband. This patient was referred to the department of obstetrics and gynecology of the Douala General Hospital which is a tertiary care centre in Cameroon after an unplanned delivery by assistant nurses at the Edea District Hospital which is a primary care centre of a fetus with hydrocephalus at breech presentation with retention of the decapitated fetal head. Her medical records showed a poor prenatal follow-up of the pregnancy. She had sought medical attention only once throughout pregnancy, at about 8 weeks gestation because of vaginal bleeding and was advised bed rest and a prescription of progesterone pessaries. No prenatal investigations were carried-out or ultrasonographic scanning performed throughout pregnancy. There is no family history of delivery of malformed fetuses. However, there was history suggestive of alcohol intake during this pregnancy but the quantity consumed per day could not be evaluated with certainty. Furthermore, her medical records described macroscopic evidence of lower limbs deformities of the decapitated fetus including spina bifida. There was no autopsy report to show any internal organ anomalies.

Her physical examination at presentation showed a patient in distress with extreme fatigue, uterine height of 26 cm and abdominal circumference of 98 cm. There were multiple vaginal lacerations including a cervical tear which had been poorly repaired. We immediately, decided on an emergency cesarean delivery of the retained decapitated fetal head and repair of the vaginal lesions under general anesthesia taking into consideration the patients' traumatized state.

Before this procedure, blood had been drawn from the patient for pre-operative studies and also investigation for possible causes of the multiple fetal anomalies. The tests requested for included toxoplasmosis, rubella, cytomegalovirus and herpes simplex virus (TORCH syndrome), full blood count, coagulation studies, HIV



Figure 1: Cesarean delivery of the decapitated fetal head.

and Hepatitis B surface antigen assays. All results were normal.

The operation performed was a lower segment transverse cesarean delivery of the decapitated retained fetal head including vaginal and cervical repair with appropriate suture materials and technique.

The post-operative stay in hospital was uneventful. Unfortunately, it was not possible to send the specimen to the pathology laboratory for analysis because of lack of consent from the family (Figure 1).

3 Discussion

Vaginal bleeding during early pregnancy is an important sign associated with about eight times the risk of malformation in newborns of this gestation compared to pregnancies without bleeding [8]. This fact alone indicates follow-up of fetal anatomy in such cases with targeted fetal neurosonograms [13]. The history of alcohol intake during pregnancy especially when heavy quantities are consumed between the 3rd and 8th week of gestation could be associated with such fetal malformations [20].

This case report emphasizes on the importance of prenatal diagnosis in obstetric practice. Hydrocephalus could be suspected clinically by an increased uterine height compared with the gestational age. Unfortunately, this was not the case with our patient who was seen by a health care provider only once throughout pregnancy at 8 weeks gestation because of vaginal bleeding. Prenatal diagnosis of fetal disorders and structural malformations is becoming increasingly important for several reasons [2, 14, 15, 17]. Approximately 3% of all pregnancies result in the delivery of an infant with a genetic disorder or birth defect. Such anomalies are also the biggest cause of infant mortality in the United States. Furthermore, minor malformations are also found in an additional 7–8% of neonates. Literature reports in the developing countries show an incidence of hydrocephalus in 1,6/1000 births Bhat and Babu 1998, 11/1000 births with neural tube defects (NTD) Kulkarni et al. 1998 [4]. Ultrasonography at 12–13 weeks gestation could show early fetal

anomalies which could be confirmed at 20–24 weeks gestation. Several recent case reports have described the use of fetal MRI between 25 and 36 weeks gestation to confirm the diagnosis of intracranial teratoma [12, 13]. Furthermore, various tests are currently in use in high risk groups. These include the triple panel test: The panel includes maternal serum alpha fetoprotein (AFP), serum beta human chorionic gonadotropin (b-HCG), and unconjugated estriol. The panel, along with maternal age, is a more sensitive (60–91%) screen for fetal aneuploidy. Maternal weight, race, and multiple pregnancies may affect the risk calculation. In a fetus with Down syndrome, b-HCG levels are elevated, and the other two levels are decreased. The triple panel can detect 57–67% of fetuses with Down syndrome in women younger than 35 years and 87% in older women [23]. Nevertheless, the triple panel tests including some fetal therapies mentioned later in this case report are not common in current obstetric practice in Cameroon. Therefore, ultrasonographic scanning is the mainstay of intrauterine diagnosis in our environment [9, 16]. Nonetheless, the case patient did not undergo ultrasonographic scanning throughout pregnancy as earlier mentioned because she never really had appropriate prenatal follow-up.

3.1 Indications for diagnostic tests [7, 19, 21]

Conditions that increase the risk of chromosomal anomaly include the following:

- Advanced maternal age > 35 years, (the most common indication).
- Previous offspring with chromosomal anomalies or other birth defects.
- Parental balanced translocation, inversion (manifests as recurrent pregnancy loss).
- Per vaginal bleeding in the 1st trimester of pregnancy.
- Suggestive fetal ultrasonographic findings.
- Positive maternal screening test findings.
- Pre-existing medical condition or history of being exposed to drugs, alcohol, medications, or infections known to be associated with congenital malformations in the fetus TORCH syndrome.
- Mendelian genetic trait in the parents.
- Molecular DNA diagnosis (cystic fibrosis, homozygous hemoglobin sickle disease [HbSS], fragile X).
- Enzymatic activities in villi, amniocytes, or both (Tay-Sachs disease, Refsum disease). From the above list of indications responsible for fetal anomalies, this patient had (acute/chronic or both) alcohol exposure throughout pregnancy and this fact could explain the presence of fetal hydrocephalus and associated birth defects especially if there had been an acute exposure between 3 and 8 weeks gestation (period of embryogenesis) [20]. All the blood studies carried-out were within normal limits.

3.2 Genetic counseling

As a prerequisite and as follow-up to prenatal diagnosis, families must be informed about the possible diagnosis, severity, prognosis, and available options for treatment and continuation of pregnancy [21].

3.3 Fetal therapy

3.3.1 Options and medical treatment

In the past two decades, the goal of prenatal diagnosis has changed from merely deciding about terminating the pregnancy to possible active intervention for improving the long-term outcome of the fetus. Recently, medical and surgical fetal therapies have emerged as options for the management of various fetal malformations.

- Termination of the pregnancy is an option for families in cases involving serious malformations that are incompatible with life. Examples of these conditions include severe chromosomal abnormalities associated with anomalies (e.g., trisomy 13), certain metabolic conditions, and anatomic defects, especially of the brain and kidneys [3].
- Elective cesarean delivery is indicated in fetal malformations that can cause dystocia or in cases in which immediate surgical correction in a sterile environment is likely to improve outcome or when successful elective delivery of an affected fetus is unlikely to be achieved by the vaginal mode; conjoined twins, large omphaloceles, severe hydrocephalus especially when associated with breech presentation, ruptured meningocele, large sacrococcygeal teratoma, and large cystic hygroma are some examples [3].
- Preterm delivery is indicated in a fetal disorder in which continued gestation would adversely affect the function of the involved organ system or threaten viability of the fetus. Early correction after delivery should be available and be of proven benefit to the fetus. Urinary tract obstruction, hydronephrosis, progressive hydrocephalus, volvulus or meconium ileus causing intestinal ischemia, and hydrops fetalis with growth retardation are examples of such lesions [6].
- Prenatal medical treatment is beneficial in a variety of diseases. With the availability of easier and safer access to the fetus, deficient nutrients, hormones, and substrates can be provided, and certain blocked metabolic pathways may be bypassed. Prior to selecting a case for fetal therapy, the fetal anomaly should be correctly diagnosed and its severity assessed. The benefits of treatment should outweigh the risks if left untreated and the risks of the procedure itself to the fetus and mother. Other serious malformations should be excluded and pulmonary maturity ascertained prior

to delivery, and adequate follow-up care should be provided after the intervention [5].

3.3.2 Fetal surgical procedures

Ventriculoamniotic shunts used for the decompression of obstructive hydrocephalus have had poor results and have caused procedure-related complications. Thus, their use is not indicated. Surgical procedures which may be explored include: [5, 11, 22].

- Fetal surgical procedures, both open and endoscopic, have been performed to repair myelomeningocele in utero. The open procedure is performed at 24–30 weeks' gestation and is shown to reduce both hindbrain herniation and the number of patients requiring shunts for hydrocephalus postnatally.
- An endoscopic procedure has been performed by the Vanderbilt group, which consists of maternal laparotomy, followed by placement of a split-thickness maternal skin graft over the exposed spinal cord and neural elements of the fetus. The skin graft is attached with fibrin glue prepared from autologous maternal cryoprecipitate. The procedure has been performed at 22–24 weeks' gestation, with the rationale that neurologic injury is partly acquired through exposure of neural elements to amniotic fluid and the uterine wall.

Most of the above mentioned procedures are noted for academic purposes and cannot be carried out in low-income developing countries. Nevertheless, it is worth recommending that hospitals (or medical practitioners) in this setting should know that the cornerstone to appropriate and prompt medical intervention is based in early medical evacuations from primary and secondary to tertiary care centres. We would discourage procedures (like uncontrollable traction on an on-coming head at breech presentation or excessive fundal pressure the so-called Kristeller maneuver) that could be dangerous or cause complications like uterine rupture, brachial plexus lesions or fetal head decapitation as found in the case patient.

If this patient was followed-up appropriately, the diagnosis of multiple malformations would have been made early in pregnancy before fetal viability and a decision taken to terminate the pregnancy. On the other hand, if the patient was referred to a tertiary care center, the right presentation would have been diagnosed clinically and confirmed by ultrasonographic scanning. This patient would have then benefited more from an elective cesarean delivery and avoided an inadvertent obstructed and traumatic breech delivery and the awful experience of fetal head decapitation [6]. Cesarean delivery of the retained decapitated fetal head also enabled us to evaluate the entire genitourinary system for other lesions that could not be apparent on clinical examination of this patient. In our opinion, procedures

like cephalocentesis (puncture and draining of the skull contents) would not have been appropriate.

4 Conclusion

The management of patients with malformed fetuses is supposed to have a multi-disciplinary approach involving the obstetrician, geneticist, psychologist, pediatrician trained in neonatal care, neurosurgeons and the anesthesiologist etc. Ultrasonographic scanning coupled with good clinical judgment remains the mainstay of obstetric practice in such cases here in Cameroon. The various treatment options of intrauterine fetal therapy available in developed countries cannot be carried out in our context. Nevertheless, in cases of isolated hydrocephalus, ventriculo-peritoneal shunts are currently being undertaken after birth by neurosurgeons here in Cameroon. Finally, the use of folic acid 5 mg or 400 microgram daily three months prior to gestation and during the first trimester of pregnancy has been reported to be beneficial in the prevention of neural tube defects [18]. We therefore advise this preventive modality (5 mg or 400 μ g folic acid three months before pregnancy and the first trimester of pregnancy) to Cameroonian women of child bearing age.

5 Recommendations

Basic reproductive health care services in Cameroon should be used to reduce the impact of birth defects by providing:

- Effective family planning.
- Education for couples on avoidable risks for birth defects.
- Effective preconceptional and prenatal care and educational campaigns to stress on the importance of such care
- Neonatal care that permits the early detection and best care locally available for management of birth defects.
- All women of reproductive age should routinely receive 5 milligrams or 400 micrograms of folic acid per day for the reduction of neural tube defects. This daily supplementation programs should be provided for women before and during pregnancy.
- Women should be discouraged from reproducing after age 35 to minimize the risk of chromosomal birth defects such as Down syndrome.
- Women should be vaccinated against rubella before they reach reproductive age to prevent congenital rubella syndrome.
- Education programs and public health messages should counsel women to limit or avoid alcohol consumption during pregnancy particularly during the early weeks and also avoid locally available teratogens.
- Finally, the ministry of health should put in place a national registry of congenital fetal malformations in Cameroon.

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