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Acrania: Report of a Rare Congenital Malformation

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ABSTRACT

Acrania is a rare lethal congenital anomaly in which there is partial or complete absence of the fetal scalp bones (calvarium). The brain tissue is completely but abnormally developed.

We present a 22-year-old primigravida, an unmarried secondary school drop-out who presented for prenatal care at 31 weeks of gestation and developed pre-term prelabor rupture of membranes a week later. An obstetric scan at presentation revealed acrania of the fetus and she was offered medical termination of the pregnancy which was declined insisting on awaiting fetal maturity. She was managed conservatively until 34weeks of gestation when she had a lower segment caesarean section. The Outcome was a live 1.8kg female fetus with acrania and other malformations who suffered an Early Neonatal Death.

Her late antenatal booking deprived her of an early diagnosis when counselling for medical termination would have been more acceptable while her religious background gave her optimism for a favourable outcome despite adequate counselling on the prognosis for this very rare lethal medical condition diagnosed in utero.

INTRODUCTION

Acrania is a rare lethal congenital anomaly in which there is partial or complete absence of the fetal scalp bones (calvarium).[1,2] The brain tissue is completely but abnormally developed unlike in anencephaly.[2] It is frequently confused with anencephaly. Most cases are diagnosed by 1st trimester ultrasound scan.
It is extremely rare and its sporadic nature suggests a low recurrence risk.[2]Only 6 cases had been reported in the literature[1] by 1996 with 5 of them diagnosed by sonography in the first trimester and terminated electively while one is the first known surviving case`. Few other caseshave been reported since then largely not surviving beyond infancy.[3,4]

The first known surviving case was the child of a 29 year old Japanese woman delivered at 38 weeks gestation by vaginal delivery as desired by the patient despite full knowledge of the risk of fetal death.[1] The outcome was a male 2.47kg infant with Apgar scores of 4 at 1minute and 2 at 5 minutes. He had scalp and dural defect, subarachnoid haemorrhage, cerebrospinal fluid leakage and partial cerebral contusion. He was observed without resuscitation and 10mins after delivery his respiration and general condition improved. At the request of his parents he subsequently underwent repair of the scalp defect and cerebrospinal fluid leakage and at 3 months of age had a sub-duro-peritoneal shunt for hydrocephalus. He was severely retarded with a developmental quotient of 10 at 3 years.[1,3]

CASE REPORT

Ms ON is a 22 year old primigravida, an unmarried secondary school drop-out, who was registered for antenatal care in our facility at 31 weeks gestation. She was brought by her church pastors' wife who was responsible for her bills.

Her pregnancy was undesired. She attempted termination of the pregnancy at about 6 weeks GA with herbal drugs and quinine tablets. Her pregnancy had subsequently been uneventful until she presented to us for antenatal care. She had received 2 doses of tetanus toxoid from a midwife who offered her some form of antenatal care prior to presentation. Her booking parameters where a blood pressure of 90/60mmHg; weight of 56kg; blood group was O rhesus 'D' positive; haemoglobin genotype was AA; packed cell volume was 36% and she was sero-negative for hepatitis B surface antigen and HIV I&II. Her Venereal Disease Research Laboratory (VDRL) test was non-reactive. Her urine was negative for protein and glucose.

Her menarche was at 15 years of age. She menstruated for 4 days in a regular monthly cycle. She had no menstrual abnormalities. She was aware of contraception but did not practice any.She was the second child in a family of 2 children, both girls. Her mother died of an unknown illness when she was a teenager and her father was unemployed.She had no family history of chronic medical illness. She was an unmarried secondary school drop-out. She was impregnated by her teenage boyfriend who denied paternity. She denied consumption of alcoholic beverages nor tobacco products.

A week after registration for antenatal care, she presented to the labour ward, at 32 weeks gestation, with complaints of gush of fluid per vaginam of 4 hours duration. There was no associated abdominal pain nor bleeding per vaginam. There was no history of trauma or other constitutional symptoms.

On examination, the feto-maternal vital signs were within normal limits and liquor drainage was confirmed on sterile speculum examination. There was no cord prolapse. A full blood count and C-reactive protein were not suggestive of overt/occult infection.

An obstetric ultrasound scan done on admission revealed a single active fetus in utero, in longitudinal lie and cephalic presentation. The biophysical profile score was 8/12, the maturity was 32 weeks of gestation and the estimated fetal weight was 1.8kg. Organ survey revealed no gross anomaly of the cardiovascular, gastrointestinal and urogenital systems. There was however absent cranial bones, exaggerated brain matter and asymmetry of the normal spinal lordosis. There was moderate oligohydramnios. The placenta was posterio- fundal in position. The cervical os was mildly dilated. There were no co-existing masses.

A diagnosis of preterm prelabour rupture of fetal membranes with congenital malformations including Acrania was made. The diagnosis, the fetal prognosis and the need for medical termination of the pregnancy was explained to the patient but she declined consent because she was hopeful of a favourable outcome despite adequate counselling and had the active support of her pastor. She was admitted into the maternity ward, placed on strict bed rest, received prophylactic antibiotics and parenteral dexamethasone to aid lung maturity.

She was managed conservatively until 34 weeks of gestation when she was offered a lower segment Caesarean Section. Intra-operative findings were a well formed lower uterine segment;a live female 1.8kg infant in cephalic presentation, APGAR scores were 4^{1},0^{5} with multiple congenital anomalies including absent fetal skull bones, prominent brain matter, absent nose, absent forearms/fingers and prominent toe on both lower limbs; the placenta was fundal and weighed 400g; the ovaries, tubes and bladder appeared normal; there was minimal amniotic fluid and estimated blood loss was 400mls. The fetus made a few gasps of breath and died within 5 minutes of delivery.

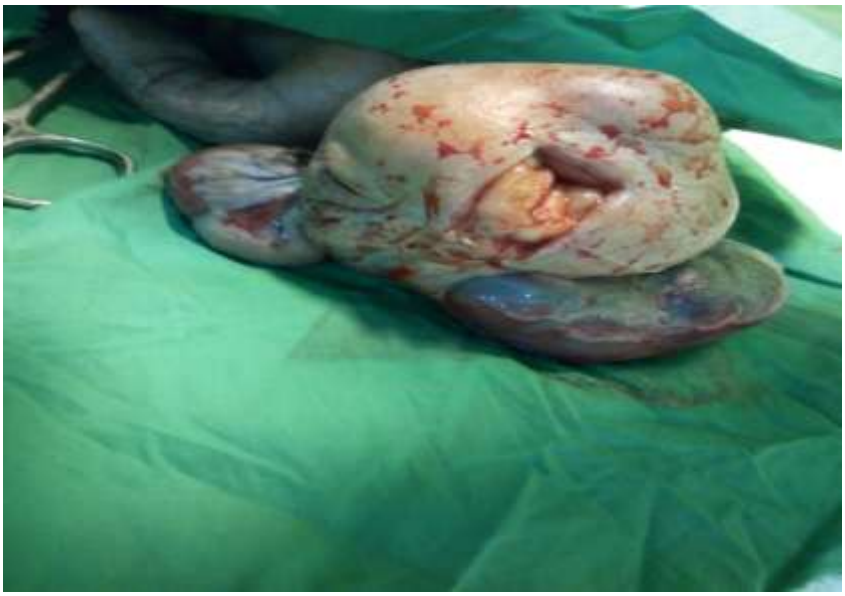

Figure 1: Cranial and Facial anomalies

Figure 2: Limb anomalies

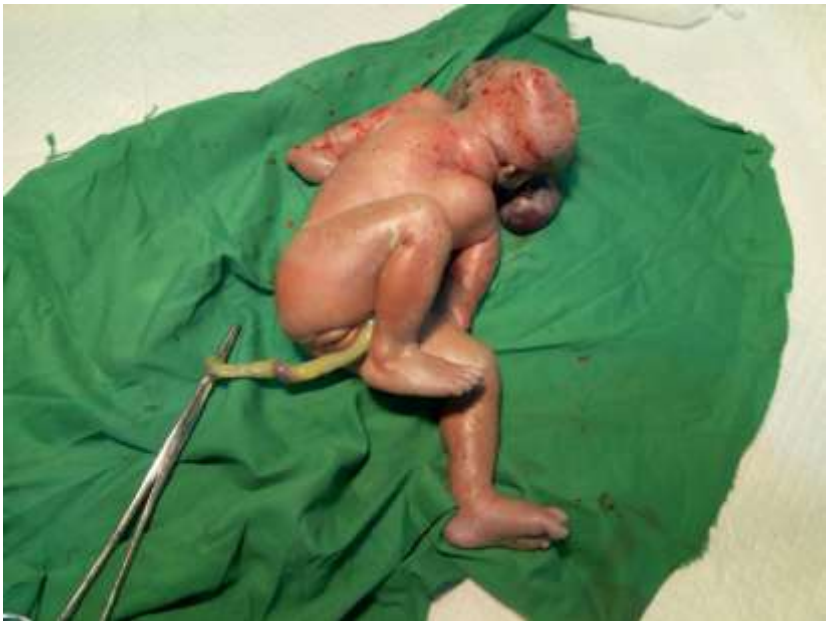

Figure 3: Lower limb anomalies

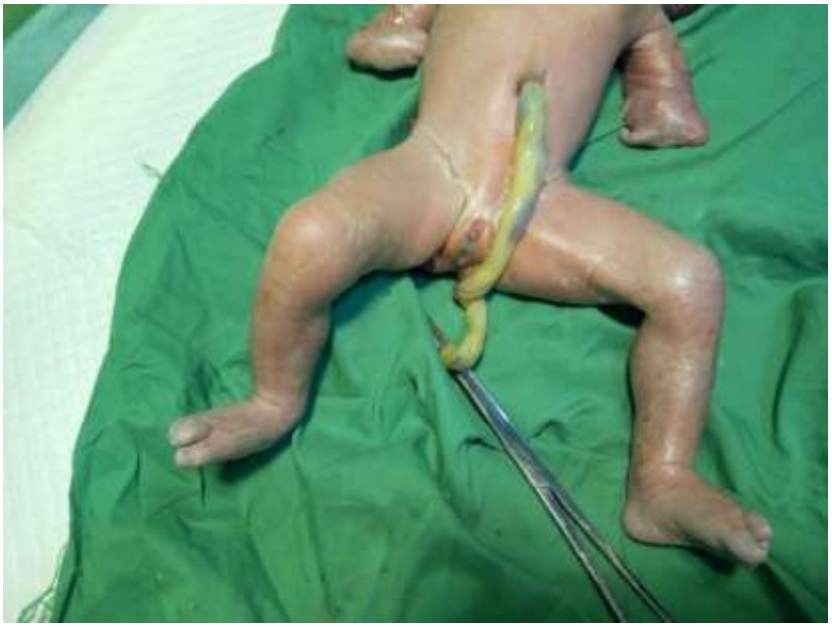

Figure 4: Genitalia

Post operatively, she received prophylactic antibiotics and analgesics. She also received intravenous 5% dextrose saline for the 1st 24 hours and thereafter commenced on graded oral sips and progressed to fluid and normal diet. Her post operative period was uneventful, she received further counselling and was discharged home on the 5th post operative day in satisfactory clinical condition. She was seen in the post natal clinic after 6 weeks and was in satisfactory clinical condition. The surgical site was healed and the uterus had involuted. She had resumed menstruation. She was further counselled on the diagnosis and family planning and was discharged from the clinic.

DISCUSSION

Acrania is extremely rare and its sporadic nature suggests a low recurrence risk.[2] Patient counselling is difficult because there is no evidence of specific genetic origin.[2] It is often difficult to distinguish between anencephaly, acrania and amniotic band sequence prenatally[5]and postnatal differentiation as was done in our patient is imperative. Amniotic band syndrome is a collection of fetal malformations associated with fibrous bands that appear to entrap or entangle various fetal parts in utero and can affect any organ or system and cause a single or multiple anomalies.[6,10]

The diagnosis of acrania can be established sonographically in the first trimester if a large mass of disorganised brain tissue covered only by a thin membrane is detected.[7,9]

Our patients' late antenatal registration which is common with teenage pregnancies excluded early diagnosis of her condition at a gestational age when medical termination of the pregnancy may have been acceptable. Her situation was further compounded by her religious background of being sponsored by her pastor who is unlikely to accept medical termination of the pregnancy. She thus ended up with a caesarean section scar for a fetus with minimal chance of survival.

Continuous advocacy onthe need to minimize out of school children, female education, provision of youth friendly contraceptive services,early antenatal registration of teenage pregnancy and re-orientation of religious bodies on medically indicated interventions will improve outcomes and avoid unnecessary surgical interventions.

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