

A CASE OF AMELIA: WHAT ARE THE IMPLICATIONS?

***J.D. SEFFAH, K.A. AMPOFO¹, E.K. SROFENYOH¹ AND I.O. KORENTENG¹**
Departments of Obstetric and Gynaecology, University of Ghana Medical School and ¹Korle Bu Teaching Hospital, Accra, Ghana.

SUMMARY

A case of Amelia (a skeletal dysplasia with failure of formation of all four limbs) is presented. The patient, aged 36, has had a previous normal baby. The index pregnancy was supervised at a polyclinic. An ultrasound examination was done to confirm the pregnancy at 11 weeks. She was referred to the Korle Bu Teaching Hospital at 42 weeks of gestation. She had a successful induction of labour and delivered a live 1.95kg male infant with no limbs. The neonatologist detected no other abnormalities. The Social Welfare department adopted him as the very depressed parents rejected him.

We advocate routine obstetric scan at 18-20 weeks gestation to detect anomalies. Apart from effective counselling, both medical and social support services should be improved for adequate and appropriate management of congenital malformations.

Keywords: Amelia, ultrasonography, management, implications

INTRODUCTION

Skeletal dysplasias are a heterogeneous group of disorders that affect the development of chondro-osseous tissues and result in abnormalities in the size and shape of different segments of the skeleton¹.

Limb buds first appear during the third week of gestation with the upper limb buds appearing a few days before the lower limb buds. Failure of formation of the limb primordia during early embryogenesis may be secondary to vascular, mechanical or teratogenic exposure.

The aetiology may be genetic and a Mendelian pattern of inheritance has been described¹. Spontaneous mutations due to environmental factors like exposure to drugs (thalidomide and warfarin), radiation, hyperthermia and hyperglycemia have also

been described. The common factor underlying all these agents may be hypoperfusion of specific areas of the embryo due to vascular disruptions. The incidence is known to be 2.4 in 10,000 births² globally but the incidence in Ghana is not known.

This is the first reported case of amelia in the Ghanaian literature and this paper seeks to highlight its occurrence and the pragmatic measures for early detection.

CASE REPORT

A 36-year-old lady, para 1, was referred to the Korle Bu Teaching Hospital (KBTH) at 42 weeks gestation because she was post-date. She had been a regular attendant at the Mamprobi Polyclinic. She had had an early ultrasound to date the pregnancy at 11 weeks gestation.

Her booking haemoglobin was 11.0g%, and the sickling test was negative. Her glucose-6-phosphate dehydrogenase (G6PD) status was normal. Her blood group was A Rhesus positive. The venereal disease research laboratory (VDRL) test for syphilis was non-reactive. Her urinalysis was normal. The fasting blood sugar (FBS) and 2-hour postprandial blood sugar level (2HPP) were within normal ranges. She had an uneventful pregnancy labour and delivery four years earlier. The child (a boy) was alive and healthy and with no congenital abnormality.

Although an asthmatic, she never had an attack throughout the pregnancy. She had no family history of diabetes, sickle cell disease, asthma, hypertension or congenital abnormalities.

She was a seamstress who was married to a driver. Both did not smoke or drink alcohol. She was prescribed routine antenatal medications of fersolate, folic acid, and weekly daraprim. She took a "lot of tea and salt for bouts of fever".

* Author for correspondence

The patient was admitted at the maternity ward of the KBTH. Her general condition was satisfactory. She was not pale or jaundiced. She had no peripheral lymphadenopathy. The thyroid gland and the breast appeared normal. She was normotensive and her chest was clinically clear.

Apart from the gravid uterus, no abnormality was detected in the abdomen. The symphysio-fundal height (SFH) was 36cm and the fetus was presenting with the vertex. The fetal heart rate was normal at 136 beats per minute. A pelvic examination showed a Bishop score of 6 (denoting the response of the cervix to induction). After counselling her about the need to induce labour at 42 weeks as well as possible complications such as ruptured uterus and foetal death, she agreed to induction of labour, which was performed, with intravaginal application of 50microgram misoprostol. Her labour was monitored with the partograph. There was no fetal distress and the induction to delivery interval was approximately 4 hours 15 minutes. The main findings at birth were;

- Live 1.95kg male infant with no limbs. There was a small appendage on the left shoulder. There was no cardio pulmonary distress. The neonatologist examined the baby further but detected no other gross anomaly.
- The placenta weighed 550g, and was delivered by controlled cord traction.
- 3 vessels were seen in the umbilical cord.
- Estimated blood loss was 400mls
- The external genitalia were well developed with 2 testes in the scrotum

The parents were very depressed and rejected the baby. After a week of admission at the maternity ward, the Social Welfare Department adopted the baby.

DISCUSSION

Amelia is the complete absence of the skeletal parts of the upper or lower limbs with no bony structure distal to the defect. Total amelia affects all four limbs. This unfortunate malformation generally is a random event, but is occasionally seen in specific syndromes associated with other congenital anomalies.

Multiple organ system defects have been associated with amelia, or limb reduction abnormalities including cardiovascular, gastrointestinal, urogenital, skeletal, neural tube, and respiratory anomalies.

Amelia is a rare finding and it is associated with medical, social and ethical implications. Some questions to be answered therefore were:

- Shouldn't the diagnosis have been made antenatally?
- What could have been the aetiology?
- What should have been the antenatal management?
- What should have been the subsequent management after birth?

At 36 years of age, the patient should have benefited from prenatal diagnosis employing maternal alpha-feto proteins and obstetric ultrasonography. She had an early scan at 11 weeks gestation, which confirmed intrauterine pregnancy and excluded any pelvic pathology. She should have however, had another ultrasound examination at about 18 to 20 weeks of pregnancy because examination at this gestation should be able to pick up any gross anomaly such as anencephaly, achondroplasia, hydrocephalus, amelia, phocomelia³. That the patient had only one sonographic examination at 11 weeks shows that the useful technology is not being used adequately and efficiently. Sonography has also a value in the mother's autonomy. Because of its pictorial nature, it affords the ease of disclosure by the physician to the patient adequate information about the fetus and its management; it simplifies the information on the fetus; and therefore further generates voluntary decision by her to authorise or refuse a clinical management. Sonography further helps in humanizing care⁴, by making antenatal supervision pleasant.

There is no literature support that "tea and salt" combination is useful in treating febrile diseases. Such self-administration with concoctions should not be tolerated in pregnancy because one must be wary of teratogens, which may not be as well documented as thalidomide or warfarin⁵.

The differential diagnosis of amelia include phocomelia in which extremities resemble those of seal since the intervening arms and legs are absent; the Robert's syndrome (phocomelia with facial clefting defects or hypoplastic nasal alae); and the Crebe's syndrome¹ (autosomal recessive condition, described in the inbred Indian tribes of Brazil, characterized by marked hypomelia of upper and lower limbs, increasing in severity from proximal to distal segments – in contrast to Robert's syndrome, the lower limbs are more affected than the upper extremities).

Although the teratogenic potential of thalidomide has been well documented, the spontaneous occurrence of amelia and limb reduction defects in the general population is rare. Most cases have no specific aetiology, but some are seen in association

with genetically transmitted disorders such as Robert's syndrome.

The current case, which had no limbs at all, could have developed as a result of spontaneous mutation. If the diagnosis had been made antenatally by ultrasound scan at 18-20 weeks, the patient would have had the best option of management. After counselling she could have had a termination of pregnancy, which is legal in this circumstance⁶, or she could have decided to carry on with the pregnancy and delivery. Neither could have resulted in such serious emotional outbursts; depression and rejection.

Prenatal diagnosis including ultrasound examinations around 18-20 weeks of pregnancy to exclude gross fetal anomalies should be encouraged in all obstetric practice⁷. Pregnant women should always be educated to avoid unproven medications including herbal concoctions, which may be injurious to the development of the fetus.

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