

# Prenatal screening for congenital malformations: diagnosis and management in low and medium income countries

Ipyana H. Mwampagatwa<sup>a</sup> and James Charles<sup>b</sup>

<sup>a</sup> School of Medicine, University of Dodoma, Central Tanzania.

<sup>b</sup> Municipal Medical Officer of Health, Dodoma Municipal Council, Central Tanzania.

Correspondence to: Ipyana H. Mwampagatwa [imwampagatwa@yahoo.com](mailto:imwampagatwa@yahoo.com)

Prenatal screening for foetal malformation is an important component of prenatal care. Unfortunately it has not been adequately addressed in many low and medium income countries (LMIC). The literature indicates that this condition is one of the most important causes of newborn deaths.

There are many screening methods but most are expensive and some require intensive training. A relatively cheap, safe and easy-to-perform method of ultrasound can help identify more than 50% of foetal malformations. Unfortunately this technique is not readily available and even when a diagnosis is made the option of pregnancy termination involves cultural and ethical issues.

This case series attempts to address the challenges faced by LMIC care providers and clients.

**KEY WORDS:** *foetal malformation, newborn deaths, prenatal care, pregnancy termination*

## Introduction

Major congenital malformations incompatible with life occur in 2-3% of all foetuses [1]. They are the cause of 20 – 30% of perinatal deaths [2]. Apart from occasional case reports little has been published on this topic in Tanzania.

Detection and management of these malformations rely on antenatal screening. The advantages of such screening include helping the clinician and parents to manage the pregnancy including whether to continue until term, determine the likely outcome, plan for complications during labour, and after birth, and identify risk factors for future pregnancies [3]. Simply put, prenatal screening and diagnosis serves to increase the possibility of optimal care of the pregnancy in terms of antenatal care, and referral for birth as required [1].

Many screening and diagnostic procedures exist (both non-invasive and invasive). Ultrasound scanning is non-invasive but sometimes a combination of techniques are needed to increase the accuracy of diagnosis [4]. This article shows the dilemma that clinicians face in making decisions in resource constrained settings.

## Material and methods

Randomly selected congenitally malformed babies born at Makole Municipal Hospital (2011 - 2014) were photographed and only represent a proportion of all the malformed babies born here.

The Facility is a District Hospital run by the Dodoma Municipal Council with a catchment population of approximately 450,000 [5]. The facility provides antenatal services and delivers about 30 babies daily.

## Case reports

The following cases were attending antenatal clinics in different facilities and referred for delivery at this hospital because of suspected abnormalities.

**Case 1.** This baby was born vaginally at 36 weeks to a primigravida woman aged 23 years – see Figure 1. The only reported finding antenatally was an abnormal lie. Ultrasound had not been available. The baby had an Apgar score 5 at birth and 1 at five minutes and died on the same day.



Figure 1. Case 1 (credit Municipal Medical officer, Dodoma)

**Case 2.** This anencephalic baby was stillborn vaginally at 37 weeks gestation to a primigravida aged 27 years – see Figure 2. The diagnosis of anencephaly was made by ultrasound at 23 weeks but the pregnancy continued to term. There was no history of drugs use or alcohol consumption



Figure 2. Case 2 (credit Municipal Medical officer, Dodoma)

**Case 3.** This stillborn baby had anencephaly and abdominal wall defects and delivered at 30 weeks to a 26 year old gravida 3 – see Figure 3. Apart from a low fundal height compared to dates, no abnormality was noted. Ultrasound had not been available.



Figure 3. Case 3 (credit Municipal Medical officer, Dodoma)

**Case 4.** This baby with anencephaly and abdominal wall defects was delivered alive by a 30 years old gravida 2 but died hours later – see Figure 4. A twin pregnancy had been suspected but ultrasound had not been available.



Figure 4. Case 4 (credit Municipal Medical officer, Dodoma)

## Discussion

Studies have shown that 70-92% of the central nervous and cardiovascular abnormalities occur in women with low risk [6]. This indicates the importance of screening of all pregnancies even if they fall in the low risk category.

The lack of appropriate affordable diagnostic equipment and expertise contributes to low detection rates in low and medium income countries (LMIC). Techniques for screening fall into two groups:

- Non-invasive: biochemical markers, isolation of foetal cells in maternal circulation, and ultrasound.
- Invasive: amniocentesis, chorionic villi sampling, foetal blood and tissue sampling.

Ultrasound is a simple and affordable method [7] but was not available in three of the cases described here. If ultrasound and trained personnel had been available, the malformations could have been detected earlier with subsequent more appropriate management – including counseling of the parents to reduce the grief and self-blame.

The accuracy of ultrasound varies from 22% to 74% [7]. This depends on the ultrasonographic skill, gestation age and type of malformation. However it remains the most affordable and low risk technique. There is a need for policy makers to introduce guidelines for screening using this approach.

Management is an enormous challenge and falls into three categories:

- Continue with the pregnancy.
- Foetal treatment where feasible.
- Termination of pregnancy.

For most LMIC, foetal termination of malformed fetuses is not an option. In Tanzania, for example, the only legal indication for termination is when the life of the mother is endangered. Therefore, whatever the prenatal findings, continuing with the pregnancy is the only option [8].

In the USA, most (95%) detected severely malformed foetuses are electively aborted [9]. These observations suggest that a review of the law in Tanzania should be considered to allow severely malformed babies incompatible with life to be electively aborted.

Ethical issues also arise when it is known that a malformation is incompatible with life. Faced with such a dilemma parents are intensely stressed [10]. In Case 2, the couple opted to continue to term but were severely disappointed at birth. Ethical principles also put clinicians in a dilemma and pose a challenge in deciding whether or not to terminate the pregnancy. Some clinicians take the

view that termination is morally unjustified while others advocate termination when the foetus is felt unlikely to survive [11].

Another challenge that face LMIC is the accuracy of the ultrasound diagnosis. In the developed world other multiple methods, including ultrasound, are combined to make a clearer diagnosis [12].

In many LMIC documentation of the incidence of congenital malformations, miscarriages and deaths is inadequate and therefore the available data may not be reliable and the magnitude of the problem cannot be adequately addressed. In developed countries like the United Kingdom congenital malformation are recorded hence their data are highly reliable [13].

The literature indicates that about 20% of the malformations are severe and life threatening [14]. Care of the surviving babies is always costly and frequently lifelong [15] which LMIC cannot afford; these babies eventually die distressing both parents and practitioners.

### Conclusion

Prenatal diagnosis and management of foetal malformations in developing countries remain a challenge. There is a need for rational guidelines adapted to local circumstances, adequate diagnostic equipment and trained personnel. Viewed from the practitioner's perspective, the abortion law in Tanzania may need review.

### Ethical issues:

Mothers gave informed consent to take photographs of their babies and authorization for publication was obtained from the Facility Management. Ethical clearance was obtained from the University of Dodoma Ethical Committee.

**Conflicts of interest:** None

### References

- Garne E, Loane M, Dolk H, De Vigan, et al. Prenatal diagnosis of severe structural congenital malformations in Europe. *Ultrasound Obstet Gynecol* 2005;25:6-11
- Shama Munim, Salva Nadeem Nadya Ali Khuwaja. Accuracy of ultrasound in the diagnosis of congenital abnormalities. *JPMA* 56:16;2006:1-5
- Schoonen M, van der Zee B, Wildschut H, de Beaufort I, de Wert G, de Koning H, et al. Informing on prenatal screening for Down syndrome prior to conception. An empirical and ethical perspective. *Am J Med Genet A*. 2012;158A:485-497
- Phillips OP, Elias S, Shulman LP, Andersen RN, Morgan CD, Simpson JL. Maternal serum screening for fetal Down syndrome in women less than 35 years of age using alpha-fetoprotein, hCG, and unconjugated estriol: a prospective 2-year study. *Obstet Gynecol*. Sep 1992;80(3 Pt 1):353-8
- National Bureau of Statistics (NBS) [Tanzania] and ICF Macro. 2011. Tanzania Demographic and Health Survey 2010. Dar es Salaam, Tanzania: NBS and ICF Macro
- Todros T, Capuzzo E, Gaglioti P. Prenatal Diagnosis of Congenital diagnosis. *Images Paediatr Cardiol* 2001;3(2):3-18
- Luck CA. The value of routine ultrasound scanning at 19 weeks: a four year study of 8849 deliveries. *Br Med J* 1992;304:1474-8.
- Tanzania Penal Code, Chapter 16 of the Laws (Revised), Issued Under Cap. 1, s. 18, <[http://www.un.org/Depts/los/LEGISLATIONANDTREATIES/PDFFILES/TZA\\_penal\\_code.pdf](http://www.un.org/Depts/los/LEGISLATIONANDTREATIES/PDFFILES/TZA_penal_code.pdf)>, accessed Nov. 26, 2012.
- Shewmon, DA et al. The use of anencephalic infants as organ sources: a critique. *Journal of the American Medical Association* 1989;261(12): 1773-1781.
- Bins V, Hsu N. 2002. Prenatal Diagnosis. Encyclopedia of life Sciences. Macmillan Publishers Ltd, Nature Publishing Group/www.els.net
- Cutter MAG. Moral pluralism and the use of anencephalic tissue and organs. *Journal of Medicine and Philosophy*, 1989, 14(1): 89-95.
- Newton Garner S. Disease registers in England. Oxford: Institute of Health Sciences, University of Oxford. 2002. ISBN 1 8407 5028
- EUROCAT 2010, 'EUROCAT Special report: Prenatal screening policies in Europe (2010)' EUROCAT Central Registry, University of Ulster.
- Lynberg MC, Edmonds LD. Surveillance of birth defects. In Halperin W. Baker EL, eds: Public Health Surveillance, New York: Van Nostrand Reinhold 1992;157-77
- Queiber-Luft A, Spranger J. Congenital Malformations. *Dtsch Arztebl* 2006;103 (38):A 2461-71. Available at [www.aerzteblatt.de](http://www.aerzteblatt.de)

Many thanks to Emma Blake, Richard Lewis, Nancy MacKeith, Tim Mellor and Simon D Taylor-Robinson for helping to prepare this issue, and to everyone who replied to our call for new reviewers.