

Sirenomelia: a Case Report

T Swende, P Abata

Citation

T Swende, P Abata. *Sirenomelia: a Case Report*. The Internet Journal of Gynecology and Obstetrics. 2009 Volume 13 Number 1.

Abstract

Sirenomelia is a rare developmental anomaly of the caudal region of the body with varying degrees of fusion of the lower limbs. It bears resemblance to the mermaid of Greek mythology. Affected individuals exhibit a variable range of associated defects. A case of sirenomelia is presented with associated absent external genitalia, urogenital and anal orifices. The precise aetiology of sirenomelia is not known but many theories have been proposed. Antenatal diagnosis of this universally lethal condition is desirable so that possible termination of pregnancy can be offered at the earliest. A good understanding of this rare condition remains central to effective counseling and management of affected couples.

INTRODUCTION

Sirenomelia, an anomalous development of the caudal region of the body, with varying degrees of fusion of the lower limbs has attracted the attention of the medical world for the past 450 years.¹ This deformity bears resemblance to the mermaid of Greek mythology and is also known as symmelia, symposia, sympus, uromelia and monopodia.² The incidence of this rare anomaly is approximately 1/100,000 births.³ Affected individuals exhibit a variable range of defects, including hypoplasia and fusion of lower limbs, vertebral abnormalities, renal agenesis, imperforate anus, and anomalies of the genital organs.⁴ We report here a case of sirenomelia associated with other anomalies and discuss some of the theories of aetiopathogenesis.

(left foot rudimentary with two digits and right foot with five digits) normal upper limbs and face, absence of external genitalia, urogenital and anal orifices. There was no obvious vertebral column defect (Figure I). Further radiological evaluation and autopsy was not done due to cultural constraints. A diagnosis of sirenomelia associated with absent external genitalia, urogenital and anal orifices was made.

CASE REPORT

A twenty four year old gravida 2 para 0 + 1 was referred from a peripheral clinic with a history of failed induction of labour at term. The pregnancy was unbooked and she had not undergone any ultrasound examination. There was no history of medication in the first trimester but was treated for malaria twice in the second trimester with oral sulphadoxine / pyrimethamine. She had a previous spontaneous abortion at eight weeks gestation. There was no clinical or biochemical evidence of diabetes. She was not consanguineously married. The patient was delivered by emergency caesarean section of a baby with indeterminate sex and Apgar scores of 2/10 and 0/10 at one and five minutes respectively. The baby weighed 2.8 kg. Physical examination of the baby showed complete fusion of non-rotated lower limbs with fused feet

Figure 1

FIGURE I (POSTERIOR AND ANTERIOR VIEWS)



DISCUSSION

The precise aetiology of sirenomelia is not well understood. Many theories have been proposed but none of these is considered conclusive.² Sirenomelia rarely occurs as an isolated anomaly¹ Vertebral defects, neural tube defects, pelvic bone defects, genitourinary anomalies and anorectal malformations accompany most cases.⁵ These heterotopic defects in multiple congenital anomalies can be better understood by the recent concept of primary developmental defect during blastogenesis. Blastogenesis is the period in embryonal development extending from the time of fecundation to the end of gastrulation (4th week). It is during this period that the undifferentiated regions of the embryo (primary field) are differentiated to the areas of specific

morphogenetic fates (progenitor fields).¹ An insult during this period may produce defects in different progenitor fields producing polytopic manifestations. The heterogeneity depends on the timing and intensity of this insult.⁶ The insult could be teratogenic, vascular or genetic. Hibelink et al⁷ demonstrated that the intravenous administration of cadmium and lead could produce sirenomelia in the golden hamster. Stevenson et al⁸ reported that in each of the eleven sirenomelia specimens, a large artery arose from the aorta high in the abdomen beyond which the aorta and its branches were hypoplastic. They inferred that the vitelline artery steals blood and nutrients away from the caudal part of the embryo and proposed the vitelline artery steal theory. Maternal diabetes and gene defects have also been proposed as possible causative factors.⁴ Antenatal diagnosis of sirenomelia on ultrasound is possible by demonstrating the fused femur, decreased distance between two femur and decreased or absent mobility of the two limbs with respect to each other. Antenatal diagnosis of this universally lethal condition is desirable so that possible termination of pregnancy can be offered at the earliest.² The limiting factors in antenatal diagnosis especially in low resource settings would include cost, availability of ultrasound facility and expertise. A good understanding of this rare condition remains central to effective counseling and management of affected couples.

References

1. Kulkarni M.L. Abdul Manaf K.M, Prasannakumar D.G, Kulkarni Preethi M. Sirenomelia with radial dysplasia. The Indian Journal of Paediatrics. 2004; 5:447-449.
2. Taori K.B, Mitra K, Ghonge N.P, Gandhi R.O, Mammen T, Sahu J. Sirenomelia Sequence (Mermaid). Report of three cases. Ind. J. Raiol Imag. 2002; 12: 3: 399-401
3. Kallen B, Castilla E.E, Lancaster P.A, Mutchinick O, Knudsen L.B, Martinez-Frias M.L et al . The Cyclops and the Mermaid: an epidemiological study of two types of rare malformation. J.Med.Genet. 1992; 29:30-35
4. W. Langman's Medical Embryology. 9th edition. Philadelphia. Lippincott Williams and Wilkins. 2004; 79
5. Buyse M.L. Birth Defects Encyclopedia. Dover, Centre for birth defects information services, inc. 1990: 1541-1542
6. L. et al. Clinical and epidemiological study of blastogenetic anomalies and their relationship to different MCA patterns. Am.J. Med. Genet. 1997.70:11-15.
7. Hibelink DR, Kaplan. Sirenomelia: analysis in the cadmium and lead treated golden hamster. Teratology carcinog 1986; 6:431-440
8. E, Johnes K.L, Phelan M.C, Jones M.C, Barr M, Clericuzio C, et al. Vascular steal: the pathogenetic mechanism producing sirenomelia and associated defects of the viscera and soft tissues. Pediatrics 1986; 78:451-457.

Author Information

Terrumun Z. Swende, MBBS, FWACS

Department of Obstetrics and Gynaecology, Federal Medical Centre (F.M.C.) Makurdi, Nigeria.

Pine P. Abata, MBBS

Department of Obstetrics and Gynaecology, Jos University Teaching Hospital (JUTH) Jos, Nigeria.