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Sirenomelia in one of Nigerian monochorionic twins

AMO Shonubi, BA Salami, *AG Falade and *AB Ejiwunmi
Olabisi Onabanjo University Teaching Hospital, Shagamu, Department of Peadiatric, University College Hospital, Ibadan, *Department of Anatomy, Olabisi Onabanjo University, Ago-Iwoye, Nigeria*

Summary
A 1,600gm baby with sirenomelia (caudal regression syndrome) associated with extensive anomalies in the internal organs occurring in one of a set of monochromic twins delivered at the Olabisi Onabanjo University Teaching Hospital (OOUTH), Shagamu, Nigeria is being reported. The baby lived for approximately twenty hours. The co-twin had no obvious malformation.

Keywords: *Sirenomelia, caudal regression syndrome, multiple congenital malformations.*

Résumé
Une bébé de 1600 grammes avec la seronemelie (syndrome de régression caudale) associe d'anormalies intense des organes interne apparut dans un groupe de jumeaux identiques accouchés a' l'hopital d'Olabisi Onabanjo (OOUTH), Shagamu, Nigeria. Le bébé avait vécu approroximativement pendant vingt heures, le second jumeau n'avait aucune malformation remarquable.

Introduction

Sirenomelia, a complex malformation first described by Duhamel in 1961 [1] is one of the most interesting conditions in teratology. With a reported incidence of about 1.5-4.2/100,000 live births [2], the abnormality had not been reported from Nigeria as shown by perusal through the literature. It is known by such terms as caudal regression syndrome and the 'mermaid'. The main feature is the presence of a single lower limb with the absence of anal and genital structures. Sirenomelia arouses further interest because of the associated abnormalities in the vertebral, anal, cardiac, tracheal, esophageal, renal and limb, the so-called VACTERL association.

The single lower limb may be characterized by two feet (simpus dipus), one foot (simpus monopus), or no recognizable foot (simpus apus). The deformed limb may be oriented posteriorly [3] or anteriorly [4]. In a combined series of 134 cases of sirenomelia reported by Duncan *et al* [5] and that of Stocker and Hefetz [2], the most commonly occurring associated abnormalities were; vertebral, sacrum and pelvic (100%), lower limb (100%), anorectal (97%), renal (93%) and genital (85%).

The condition is ubiquitous but very little is known with certainty about the actiology. The theory of the vitelline arterial steal [6] as evidenced by a single umbilical artery had been widely accepted as involved in its aetiology. However, because not all cases are associated with this finding (the above series reported only 79% with arterial steal), other aetiological factors proposed have included excessive cell death in the primi tive streak and hindgut mesoderm of the foetus during the critical period of development in- utero [7,8]. Genetic factors [9] and the role of teratogenic chemicals [7,8] following the thalidomide tragedy are also considered as causes. Furthermore, diabetes mellitus in the mother had been implicated in this condition as well.[1,10]

We report below an interesting case of sirenomelia born to healthy Nigerian parents from the South-western part of the country. The case is particularly interesting because only one of a set of monochoronic twins had the abnormality and it was associated with the viteline arterial steal.

Case report

A twenty-two year old, gravida 4, para 3'1 woman went into spontaneous labour and had normal vaginal delivery of a set of twins at a gestational age of 35 weeks. The mother had antenatal booking at 18 weeks gestation. At the next antenatal visit 4 weeks later, it was noticed that the fundal height was bigger than the gestational age of pregnancy. An ultrasound scan at 29 weeks gestation confirmed monochorionic twins with no discernible foetal abnormality. There was polyhydramnios and the estimated gestational age was 28.5±1 weeks.

A review of the obstetric and gynaecological history revealed no congenital abnormalities in any of her previous children. She denied any form of illness or ingestion of drugs (including native concoctions) other than the routine haematinics and antimalarials prescribed by her attending doctor during pregnancy.

The first twin (Baby A) was a male baby who presented with vertex, had a normal vaginal delivery with Apgar scores of 8 and 10 at 1 and 5 minutes respectively. He weighed 2,450gm and was normal. The second twin (Baby B), delivered by breech, had Apgar scores of 2 and 5 at 1 and 5 minutes respectively. He weighed 1,600gm and had gross multiple congenital malformations. There was a single placenta, which weighed 1,080gm. Baby B was centrally cyanosed in spite of adequate respiratory efforts. The occipito-frontal circumference (OFC) was 32 (-2.5D) cm and crown-heel length was 35cm (-2.5D). The baby remained centrally cyanosed despite satisfactory resuscitative measures. The respiratory rate was 52/minute with fair air entry into both lungs and coarse crepitations. The heart rate, 136/minute, was regular with normal first and second heart sounds. No murmur was heard. There was no external genitalia or anal opening. A tapering stump which was anteriorily flexed replaced the lower limbs. The baby cried poorly although no obvious neural tube defect was noted. Spontaneous movements were observed in both upper limbs and the lower single stump.

A baby-graph (Figs. 1a and 1b) revealed deformed sacrum and pelvic bone, a single femur and a rudimentary tibia. The abdominal gas pattern appeared within normal limits but did not extend to the anal verge. The baby lived for only twenty hours.

A detailed post mortem examination revealed the following extensive anomalies: high arched palate, high ano-rectal agenesis (blind ending sigmoid colon); vesical agenesis with bilateral agenesis of kidneys and ureters, bilateral undescended

Correspondence: Dr. A.M.O. Shonubi, P.O. Box 111, Shagamu, Ogun State, Nigeria

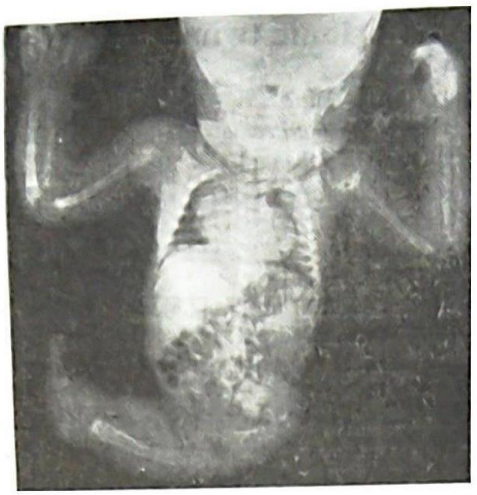

Fig. 1a: Antero-posterior baby-graph showing deformed sacrum, pelvis, single femur, rudimentary tibia and absent fibula in the simpus apus.

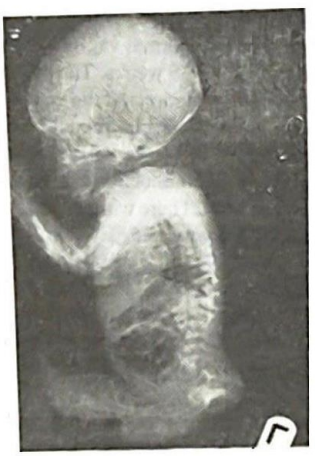

Fig. 1b: Lateral baby-graph showing the sacrum pointed posteriorly, the simpus apus anteriorly oriented and held at fixed flexion relative to the trunk

gonads; transversely lying heart with triscupid valve atresia and hypertrophic right ventricle, huge superior vena cava, a fibrotic ductus arteriosus, and a single umbilical artery originating from the aorta near the celiac axis; absent cauda equina, deformed pelvic bone, single femur and rudimentary tibia.

Discussion

The caudal regression syndrome includes a spectrum of anomalies, from imperforate anus to the extreme malformation of Sirenomelia [1,11,12]. Sirenomelia is a rare congenital malformation and its occurrence in one of a set of twins is even more rare [13,14]. Our patient who had ano-rectal agenesis, absent external genitalia, a single lower extremity and extensive anomalies of the internal organs represents a typical case of Sirenomelia [11,15].

The exact cause of Sirenomelia is still unknown. The results of the work by Chandebois and Brunet [16] suggest that the various developmental defects seem to have a single common origin viz the speeding up of the progression of cell differentiation in the notochord anlarge (which is the organisation centre of the embryo) during the regression of the Hensen's node. A gene mutation as well as mechanical stress have been incriminated in the abnormal development of the notochord anlarge [16]. The co-existence of genito-urinary, gastro-intestinal and skeletal anomalies in some of the reported cases of sirenomelia are thought to result from defects in the posterior axis mesoderm during the third or fourth week of gestation [11,12,15].

Dissection of abdominal vasculature in eleven cases of sirenomelia by Stevenson *et al* [6] has demonstrated a pattern of vascular abnormalities that explains the defects usually found in this condition. The common feature is the presence of a single large artery, arising high on the abdominal aorta, which assumes the function of the two umbilical arteries. This large artery, called "steal vessel" diverts nutrients away from the caudal end of the embryo. Thus, organs supplied by the aorta distal to the level of this steal vessel are underdeveloped, malformed or arrested in some incomplete stage [6]. In contrast to these various concepts [6,16], the single lower extremity in sirenomelia has also been attributed to failure of the lower limb bud to be cleaved into two lateral masses by an intervening allantois.[6].

Although, we did not perform whole-body angiograph (on our patient) as in the study by Talamo *et al* [17], our findings on detailed dissection, particularly that of single large umbilical artery (originating from the aorta just distal to the celiac axis), are strongly in support of the theory of "vascular steal" causing a diversion of blood flow away from pelvic structures during the critical period of development as a more likely explanation of the wide spectrum of malformations seen in our patient rather than increase in cell differentiation in the notochord anlarge during the regression of the Hensen's node.[16].

A gene mutation as well as mechanical stress resulting in the abnormal behaviour of the notochord anlarge is most unlikely in the present case in view of the fact that the first of the set of twins is normal. If the theory of gene mutation were to be applicable, both twins in a monochorionic situation would probably have manifested varying degrees of malformation. Indeed, to date, there is no direct evidence of a genetic basis for sirenomelia [18]. However, in accepting the" Vascular steal" theory, the inducing factors remain unexplained. Perhaps more detailed studies in future, based on appropriate experimental models could address this issue.

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