SIRENOMELIA (MERMAID)—CONGENITAL DEFORMITY

(A Case Report)

by

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The Greeks considered congenital deformities to be of divine origin. Birth of such monsters were thought to indicate a divine warning of some impending calamity. To please the Gods such monster infants and, in some cases, even the mothers of such monsters were sacrificed (Glenister, 1957).

The sympodial deformity has been recognised from very early times, and has found its place in myths and ancient legends. Legendary monster of the Lybian desert, the sciapod or shadow foot, was described by both Herodotus and Pliny. This monster used his large webbed foot as a sunshade while resting. The carving from a fourteenth century bench in the nave of Dennington Church, Suffolk, U.K., shows one such monster. The monster is shown to have a fused lower limb and a large webbed foot with 10 toes. However, unlike the true sympodial limb, the knee is shown to flex posteriorly. Even early scientific writers have leaned to imaginations and taken a little poetic licence in their descriptions of such monsters. Bartholin (1954) reported well developed mammary glands and webbed fingers in his case.

The word "Sirenomelia" has its origin in two Greek words. "Siren" in Greek mythology refers to sea nymphs, half woman and half fish, who lived in rocky isles to which they lured seafarers to their destruction by their singing. "Melos" means limb.

Although sirenomelia is a rare congenital deformity, a number of cases have been reported in the literature. Ballantyne (1904) found 116 cases reported in the literature. Kampmeier (1927), after an exhaustive study of the literature, added 52 additional cases after which, occasional case reports have appeared. Hendry and Kohler (1956) have also reviewed the literature. The anatomical and embryological aspects of sirenomelia have been described by Bearn (1960). Bain, *et al* (1960) have reported 4 such cases. Other cases have been reported from time to time (Foulks, and McMurray, 1954, Jolly, and Lamont, 1958 and Newbill, 1941).

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One such case of sirenomelia is reported here.

Case Report

Mrs. K., aged 35 years, gravida VI, para V, was admitted as an emergency case in labour to the Maternity Section, General Hospital, Pondicherry, on 29-4-68 at 1-0 p.m. The patient was 35 weeks pregnant. She delivered spontaneously an alive sirenomelic baby on 29-4-68 at 2.50 p.m. The volume of liquor amnii was not measured but there was no obvious variation from normal. Placenta and membranes were normal. Puerperium was uneventful and the patient was discharged on the 5th day. The baby was noted to be deeply asphyxiated at birth and expired on 29-4-68 at 7.00 p.m.

The patient had previously given birth to five children at term. One baby died at the age of 10 days due "tetanus" and another died at the age of 2 years due to "fever". The three living children were examined. They were healthy and had no evidence of any congenital abnormality.

There was no available history of any congenital malformation in the family. During the present pregnancy patient had not suffered from any febrile illness nor had she taken any drugs likely to result in foetal abnormality.

External examination of the baby

The baby weighed 1.950 kg. The head, face, thorax and upper limbs were normal. The nails on the fingers were reaching upto the tip. The external genitalia were absent and there was no urethral orifice. No congenital deformity of the spine could be palpated. A small dimple was seen immediately below the lower end of the vertebral column, representing the imperforate anus. Single lower limb was seen extending from the middle of the lower end of the trunk. The limb was so rotated that the sole of the foot was pointing anteriorly and the dorsum of the foot posteriorly. At the knee joint flexion was possible anteriorly. Two patellae were palpated on the posteriolateral aspect of the knee joint, one on either side. Only one bone could be palpated through the skin in the thigh and the leg. A single small foot was present with the sole directed anteriorly and only three toes were present, the middle one being fused with the left.

Radiological examination

Skeletal System: Skull bones do not show any abnormality. All the cervical vertebrae are seen with spina befida in C_6 and C_7. No abnormality is seen in the dorsal vertebrae in number and osseous centres. Spina bifida is seen in L_5. Bones of the pelvis were small, but no abnormality noticed. Single femur is seen with flattening of the lower end. Both tibiae are fused and no fibulae seen. No abnormality is seen in the bones of the upper extremity.

Viscera: Dilated stomach and coils of intestine seen. Lungs show minimal aeration, and air in the trachea is seen which is deviated to the right.

Internal Examination (Post-mortem).

Heart showed absence of interatrial septum. Lungs showed hypoplasia and were formed of two lobes on each side.

The kidneys were small disc-shaped. There were no well defined pelves or ureters and the bladder was absent. The suprarenals were large and disc-shaped. No gonads could be seen.

Large intestine ended blindly, 61 cms distal to ileo-caecal junction.

The umbilical artery was only one which seemed to be the continuation of aorta.

Discussion

Classification of Sympodial Abnormality

Forster (1861, 1865) has classified sympodial foetuses into three degrees, depending on the completeness of fusion of the two limbs.

1. *Symelia Apus:* No feet are present. Limbs are completely fused into one single limb. One femur and one tibia are present.

2. *Symelia Unipus:* One foot is present which is formed by a partial fusion of the two feet. Upto ten toes may be found. Little toes may be

fused together. Plantar surface of the foot faces anteriorly. Two femora and two tibiae and fibulae are present, the latter bone being medial to each tibia. The main muscle groups can be identified.

3. *Symelia Bipus:* Two feet are present giving the appearance of fins, hence the name "mermaid" foetus for this abnormality. The fusion of the limbs extends only as far as the ankles and the muscles of the two limbs are present.

In our case, there was one foot with three toes. There were only one femur and one tibia which seemed to have been formed by the fusion of two tibiae. No fibulae were present The interesting point was the presence of two distinct patellae.

Non-rotation of Lower Limbs

This is a constant finding in all cases of sirenomelia. The knee cap is on the back, the legs bend anteriorly and the sole of the foot is directed anteriorly. This position of the fused limbs is of embryological interest. It demonstrates the persistance of the early unrotated position of the limbs. The normal lower limb undergoes internal rotation, so that the post-axial border and digit of the little toe become lateral and the ventral surface faces posteriorly. In the sympodial deformity, the developing limb buds are fused from the start along their post-axial borders and rotation is therefore impossible.

Bain and Scott (1960) have described large flattened and clumsy appearance of hands associated with this deformity.

The Association of Sirenomelia with Potter's syndrome

Potter (1946) described a syndrome of renal agenesis, hypoplasia of the lungs and the typical facial appearance of the foetus. It is possible that the association between this syndrome and sirenomelia is common, because in this abnormality there is usually complete agenesis of renal tissues. Bearn (1960) has described a case and discussed the association. However, Jones and Lee (1955) described a case of sirenomelia with normal facies in spite of complete absence of renal tissue.

Our case did not show the typical facial appearance described by Potter. Although small disc-shaped kidneys were present, there was complete absence of pelves, ureters and the bladder. However, hypoplasia of lungs was present in our case.

The absence of the kidneys also explains the large flattened appearance of the adrenal glands. The kidneys presumably are responsible for moulding the adrenals into their characteristic shape in the normal foetus.

Single Umbilical Artery

This is a common finding in all the reported cases. The single umbilical artery connects directly with the aorta. Ballantyne (1904) and Kampmeier (1927) found that the artery within the umbilical cord in their cases was a persistant vitelline artery.

Sex of the Foetus

It would be in keeping with the popular mythical belief and in justification of the word "mermaid" used

for these foetuses if their sex was female. However, in many cases no external or internal genital organs are found. The nuclear chromatin pattern is less specific to this condition and little can be deduced from it. In all reported cases where sex could be made out there is predominance of the male sex. Kampmeier (1927) found 38 males out of a series of 52. Potter (1961) in her series of renal agenesis found only four out of 26 being females. Bearn (1960) has reported a sirenomelic foetus with male sex. Bain, *et al* (1960) have reported four cases out of which 2 were male, 1 female and in one, no internal genitalia were found.

In our case, neither external nor internal genital organs were found.

Theories of Causation

(1) References to the volume of liquor amnii are generally vague. It seems to be the impression that oligohydramnios is commonly associated with sympodial defects. Lack of amniotic fluid is regarded as the mechanical cause of minor malformation of the extremities like talipes. It is unlikely that this could cause severe degrees of malformation like sympodial defects. Perhaps, oligohydramnios is the result of the renal agenesis found in sirenomelic foetuses rather than the cause for sympodial deformity. Oligo-hydramnios may account for the typical appearance produced in the hands and face.

(2) The single umbilical artery is postulated to cause a primary vascular defect in the growing limb buds, resulting in its faulty development. There is little evidence in the support of this view. Absence of one umbilical artery is frequently seen in babies with varied congenital malformation and also, occasionally in apparently normal infants.

(3) The formation of ureters, aorta, umbilical arteries and the limbs is constantly disturbed in sirenomelia. At 4.5 mm stage of the embryo, the ureteric buds and the limb buds appear and hence the disturbing factor seems to act at this or yet early stage. The cause of these malformations could be either genetic damage or injury in the developing embryo by factors as yet unknown to us.

Bolk (1899) and Ballantyne (1904) suggested that the defect was primarily a failure of development of caudal segments at a very early stage, before the limb buds forms. Bearn (1960) has discussed in details the causation of sympodial deformity.

There is experimental evidence to support the above view. Wolff (1936) has produced the sympodial deformity in the chick by destroying the caudal end of the early embryo with x-rays.

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Figs. on Art Paper IX