Sirenomelia with Potter’s facies - a case report

Siva Sree Ranga MK, 1Arun Kumar S Bilodi, 2 Rijied Thompson Swer
1Postgraduate student, 2Professor 3Assistant Professor
Department of Anatomy, Mahatma Gandhi Medical College and Research Institute, Pilliyarkuppam,. Pondicherry

Abstract

A specimen of Sirenomelia from the department of Anatomy, Mahatma Gandhi Medical College and Research Institute, Pondicherry is presented here. This specimen was examined in detail and X-Ray was taken. On examination, the specimen showed i) Fused lower limb like a stump, ii) Undifferentiated genital organs, iii) Widely spaced nipples, iv) Receded chin and webbed neck, v) Low set large ears, vi) Epicanthal fold, vii) Flattened nose, viii) Absence of anal orifice. Fused lower limbs with associated undifferentiated genital organs and absence of anal orifice was found interesting to study and reported here as a case. The presence of Fused lower limb like a stump is suggestive of Sirenomelia and presence of i) Widely spaced nipples ii) Receded chin and webbed neck, iii) Low set large ears, v) Epicanthal fold, vi) Flattened nose is suggestive of Potter’s Facies. It is presented here due to rarity of occurrence of such anomaly.

Key words: mermaid syndrome, idiopathic etiology, congenital abnormality

Introduction

Sirenomelia is congenital abnormality which is commonly known as Mermaid syndrome involving lower limbs which are fused. There may be associated gastrointestinal or urogenital anomalies. The word is derived from sirens of Greek and in Roman mythology. This anomaly is known to occur during 28-32 days of Intra Uterine Life. Pathogenesis of this anomaly is not clear, but studies have reported that there is a causal regression or vascular steal phenomenon. The incidence of Sirenomelia is 1.05 in 1,00,000 births. It is common in males (three times), incidences are higher in identical twins and also increase of risk before the maternal age of 20 and after the elderly age of 40 years. The other associated anomalies so far reported are double inferior vena cava and Angiomatosus Lumbosacral myelocystocele.

Case Report

A full term normally delivered fetal specimen constituted the material for the present study. Exact case history of the specimen is not known. This specimen showed fusion of lower limbs with absence of anal opening but there was a small depression in the region of anal orifice.

Gross Features

i) Face, chest, upper limbs were normal ii) Fusion of lower limbs into tail like stump. iii) Undifferentiated external genital organs iv) Imperforate anus and upper limbs with normal digits but with skin changes on the dorsum of the hand and digits Other findings were: One oblique fold in mid forearm and transverse crease just above the left cubital fossa. Creases were also present over the deltoid region; Oblique folds were present in sacral region; Widely spaced nipples were seen over thoracic region; There was webbed neck and rudimentary toe with ill defined nail.

Measurement of the fetus

Head circumference - 29 cms; Length of the head - 12.5 cms; Full arm span-37 cms; Right arm length-15 cms; Left arm length span -15.5 cms; Full length of the fetus (crown to heel) - 42 cms; Chest circumference - 28cms; Abdomen circumference -27cms.

X-Ray findings

i) Lumbar Scoliosis, with convexity to the left. ii) Only left femur was present ; A part of tibia was also seen. iii) Deformed Hip bone - acetabulum was present only on left side.
Discussion

Browne et al. have reported a case of Mermaid syndrome with fusion of lower limbs associated with anomalous urogenital and gastrointestinal systems in a baby of 40 weeks weighing 1760gms. That female infant delivered normally at full term had a single lower limb with single digit; there was absence of anal orifice and external genitalia. Other findings were: hypoplasia of iliac bone on right side with agenesis of sacrum, deformities of multiple ribs, Potter’s facies, left sided ear appendage, a large lumbosacral cystic lesion which was retro peritoneal measuring 17x7x2 cms. The baby had bilateral hypoplasia of lung, proximal dilatation of colon with blind termination and absence of kidneys on both sides. Ugwu et al. reported a case of sirenomelia in 16 days old Nigerian male baby delivered normally and came with history of complete fusion of lower limbs associated with imperforate anus, neural tube defects at the level L2-L3, single umbilical artery and renal anomaly. Parents were belonging to Hausa Ethnic group and non consanguineous.

Stockert & Heifetz have classified these abnormalities into type-1 to type 7. Type-1 is least severe condition where all the bones are present where as type 7 is so severe where there is absence of tibia and fibula but presence of fused femur. Progressive oligohydraminos in the second trimester of pregnancy will be the first sign of malformation of Sirenomelia. But prenatal diagnosis is difficult. Potter’s syndrome

Fig. 1: Showing Anterior Aspect of Sirenomelia specimen having:

i) Fused lower limbs; ii) Undifferentiated genital organs; iii) Widely spaced nipples; iv) Receded chin with webbed neck; v) Low set large ears; vi) Epicanthal fold.

Fig. 2: Showing posterior aspect of Sirenomelia specimen showing:

i) Scoliosis of spine; ii) Four depressions with thick skin fold; iii) Lower limb stump showing the nail; iv) Shallow anal depression present but no anal orifice.

Fig. 3: Radiograph of museum specimen of Sirenomelia showing:

i) Lumbar scoliosis with convexity to the left; ii) Only left side femur present; iii) A part of tibia is also seen; iv) Deformed hip bone. Acetabulum present on the left side.

7 is so severe where there is absence of tibia and fibula but presence of fused femur. Progressive oligohydraminos in the second trimester of pregnancy will be the first sign of malformation of Sirenomelia. But prenatal diagnosis is difficult. Potter’s syndrome
consists of Potter’s Facies which includes large low set ears, hypertelorism, flattened nose, epicanthal folds and receded chin. In most of the cases of sirenomelia, there is single umbilical artery arising from the dorsal aortic trunk.

References


Address for communication:
Dr. Siva Sree Ranga M. K.,
Siva Sree Sadanam, Vellarada P.O.
Thiruvananthapuram Dt. - 695 505, Kerala.
e-mail ID : sivaseeranga@yahoo.com
Mobile : 09447696069