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CASE REPORT

Sirenomelia: A Case Report

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Abstract

Sirenomelia, which is also known as mermaid syndrome, is an extremely rare congenital developmental disorder characterized by anomalies of the lower spine and the lower limbs. Affected infants are born with partial or complete fusion of the legs. Additional malformations may also occur including genitourinary abnormalities, gastrointestinal abnormalities, anomalies of the lumbarsacral spine and pelvis and absence or underdevelopment (agenesis) of one or both kidneys. Affected infants may have one foot, no feet or both feet, which may be rotated externally. The tailbone is usually absent and the sacrum is partially or completely absent as well. Additional conditions may occur with sirenomelia including imperforate anus, spina bifida, and heart (cardiac) malformations. Sirenomelia is often fatal during the newborn period. : The case was reported at Zenana hospital,Berhampur ,during routine delivery.There was H/O diabetes, but no H/O of epilepsy,hypertension of the mother,after the baby was born live & it was transferred to NICU & within few hours it died . The morphology was studied & seen that the lower limbs are fused up to distal end with toes frinzizing out.There were six toes found, three in each leg. Great toe being present laterally instead of medially. Popliteal fossa was present in anterior aspect . Absense of external genitalia (sex cant be determined) . A Potter's face (ocular hypertelorism, low-set ears, receding chin and flattening of the nose)Imperforate anus . No deformity found on upperlimb, trunk, abdomen, face & head morphologically.

INTRODUCTION

Sirenomelia, which is also known as mermaid syndrome, is an extremely rare congenital developmental disorder characterized by anomalies of the lower spine and the lower limbs. Affected infants are born with partial or complete fusion of the legs. Additional malformations may also occur including genitourinary abnormalities, gastrointestinal abnormalities, anomalies of the lumbarsacral spine and pelvis and absence or underdevelopment (agenesis) of one or both kidneys. Affected infants may have one foot, no feet or both feet, which may be rotated externally. The tailbone is usually absent and the sacrum is partially or completely absent as well. Additional conditions may occur with sirenomelia including imperforate anus, spina bifida, and heart (cardiac) malformations. The condition is thought to be part of a spectrum including imperforate anus, sacral agenesis and sirenomelia[3] .Sirenomelia is often fatal during the newborn period. The exact cause of sirenomelia is unknown, most cases occur randomly for no apparent reason (sporadically). It is a lethal type of birth defect in which the two lower limbs of

the new born are attached to or fused together as one.It is other wise called as mermaid syndrome as the two fused lower limbs look like mermaid's tail. Affected newborns do not survive more than 24 hours after birth. Only four cases of a surviving infant with sirenomelia have been reported[10].

This is an uncommon congenial deformity and occurs in 1 out of every 100,000 live births in the US. The condition occurs predominantly in males, with a sex ratio of 2.7: 1[24] Infants suffering from this disorder die within a day or two due to the abnormal development of kidney and urinary bladder. Sirenomelia has been reported among different ethnic groups of the world and is common among infants born to pregnant women suffering from Diabetes mellitus.

Case Report :

The case was reported at Zenana hospital,Berhampur ,during routine delivery.There was H/O diabetes, but no H/O of epilepsy,hypertension of the mother,after the baby was born live & it was transferred to ICU & within few hours it died .Then with the permission of of the ethical committee of medical college ,the case was transferred to department of Anatomy, M.K.C.G Medical college for academic purpose ,with prior permission from H.O.D.,Anatomy,MKCGMC & Director,Zenana hospital & with consent of the parents ,

The morphology was studied (fig-1,2 &3)& seen that the lower limbs are fused up to distal end with toes frinzizing out.There are six toes found, three in each leg. Great toe being present laterally instead of medially. Popliteal fossa was present in anterior aspect . Absense of external genitalia (sex cant be determined) . A Potter's face (ocular hypertelorism, low-set ears, receding chin and flattening of the nose)Imperforate anus . No deformity found on upperlimb, trunk, abdomen, face & head morphologically.

Internal abnormalities : normal liver , renal agenesis , absence bladder found

For further study radiological study was done.X- ray of the body including the lower limb was done .X-ray findings (fig-4):both the femurs present,both tibia present but single fibula present. Partial sacral agenesis present.

Discussion :

This is an uncommon congenital deformity and occurs in 1 out of every 100,000 live births in the US. Infants suffering from this disorder die within a day or two due to the abnormal development of kidney and urinary bladder. Sirenomelia has been reported among different ethnic groups of the world and is common among infants born to pregnant women suffering from Diabetes mellitus.

The abnormalities of embryologic development leading to the sirenomelia sequence occur about the fourth week of gestation, and concern the medio-posterior mesodermic axis and the caudal blastem.[16] At this gestational age, the cesspool (cloaca) is formed, the kidneys are located in the pelvis, while the gonads are intra-abdominal. It seems logical that any damage to the caudal extremity of the embryo would affect the development of the external and internal genital organs (except the gonads, which are intra-abdominal), the terminal bowel, the bladder, the kidneys and the pelvic bones[4].

In humans, fusion of the legs in sirenomelia occurs in a spectrum of morphologies, ranging from the mildest cases in which all bones of the two fused lower limbs are discernible, to the most severe cases in which there is no indication that the single rudimentary lower limb that is present derives from the fusion of two

Infants of diabetic mothers have two to three times the average incidence of congenital anomalies. These include neural tube defects, cardiac defects (transposition of the great vessels, coarctation of the aorta, VSD, ASD, cardiomyopathy), situs inversus, renal anomalies (hydronephrosis, renal agenesis, multicystic dysplastic kidney, duplication of the renal tracts), intestinal atresia, and forms of caudal regression including sacral agenesis[17,34].

Lynch and Wright[18] reported a case where the mother had diabetes and the infant had sirenomelia with renal agenesis and an absent right radius. There was also a hypertrophic cardiomyopathy and a bicuspid pulmonary valve. Sirenomelia is thought to occur in about 1 in 65,000 live births and may be more common in one of monozygotic twins. It bears resemblance to the mermaid Greek mythology. Affected individuals exhibit a variable range of

defects, including hypoplasia and or fusion of lower limbs, vertebral abnormalities, renal agenesis, imperforate anus, and anomalies of the genital organs[27]

Several etiologic factors have been suggested; however none of them explains all the anomalies encountered in the sirenomelia sequence[4]:

Maternal diabetes is the only maternal disease known to be associated with sirenomelia (2% of cases), although this is associated more frequently with CRS (22% of cases). Teratogens: retinoic acid, cadmium and cyclophosphamide have been implicated in the genesis of sirenomelia in mice and hamsters. Genetic factors: several family cases of CRS have been reported, as well as some cases of minor vertebral abnormalities in the parents, suggesting a possible genetic transmission. Several modalities of transmission have been suggested: dominant sex-linked transmission, multifactor polygenetic transmission, and dominant autosomic transmission with a variable expression and an attenuated penetranceThe vascular steal theory: this theory was initially suggested in 1927 by Kampmeier and was called "nutritional deficit". It was reintroduced in 1986 by Stevenson[36] under the term "vascular steal". Stevenson relied on observations taken from the autopsies of 11 cases of sirenomelia, He observed a mega-artery ensures the function of both umbilical arteries and diverts the blood flow on the embryo's caudal extremity toward the placenta, causing a nutritional deficit and a lack of development of the caudal extremity. Sirenomelia is quite often accompanied by a unique umbilical artery.[37] Our observation of a single umbilical artery, renal agenesis and the absence of renal arteries might support this theory,. The gonads are vascularized, in principle, by collateral arteries of the abdominal aorta starting from the top of the renal arteries.. The vascular steal theory also fails to explain the frequent association of sirenomelia with other abnormalities such as cranial, cardiac and esophageal defects.[35]

Previous reports described the extent of the malformation complex in association with sirenomelia. M Al-Haggar et al[10] reported one case of a fetus of sirenomelia sequence with Potters syndrome which showed oligohydramnios and symelia apus. The infant showed absent urinary tract and external genitalia, the legs were fused by skin and had separate bones associated with Potter's syndrome. The mother had a history of gestational diabetes mellitus.. Anis Fadhlaoui et al[4] report a case of sirenomelia sequence observed in an incident of preterm labor during the 29th gestational week. According to some authors, this syndrome should be classified separately from caudal regression syndrome and is likely to be the result of an abnormality taking place during the fourth gestational week, causing developmental abnormalities in the lower extremities, pelvis, genitalia, urinary tract and digestive organs.McCoy et al[23] reported an unusual case with two normal kidneys. Perez-Aytes et al,[25] reported an unusual case where there appeared to be partial fusion of the lower limbs associated with absent feet, but other features of caudal regression and a single aberrant umbilical artery. Akbiyik et al.[2] reported male twins, one of which had sirenomelia and the other just an imperforate anus. Unfortunately zygosity was not established. Shonubi et al[31] reported sirenomelia in 1 of 2 monochorionic twins. Duncan et al.,[11] reviewed 445 cases ascertained for sacrococcygeal malformations and found that 12% had sirenomelia, 27% VATER association and 34% what they called the sacrococcygeal dysgenesis association Guidera et al[13] reported a case of orthopaedic abnormalities and pelvic dysplasia, fused lower extremities, and osseous fusion of the calcanei, but with normal femurs, tibias, and fibulas. This patient also had significant cardiac and renal abnormalities. Stevenson *et al*[33] reported in 1986 that the aetiology of sirenomelia was explained by vascular steal theory. They documented the common feature of a single large artery arising from high in the abdominal cavity, which they thought diverted nutrients from the caudal end of the embryo in a vascular 'steal' phenomenon. The associated anomalies are variable and involved multiple organ systems. The most common anomalies include a single umbilical artery, malformation of urinary tract, lower gastrointestinal tract and external genitalia[34].

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FIG-1

FIG-2

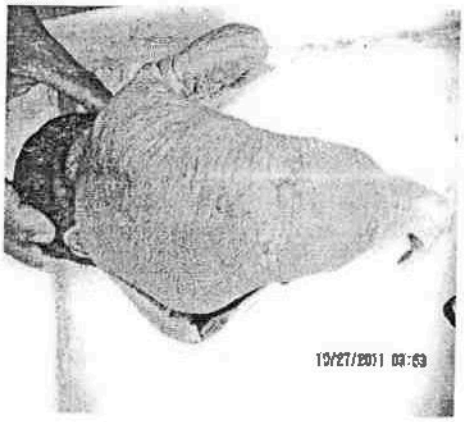

FIG -3

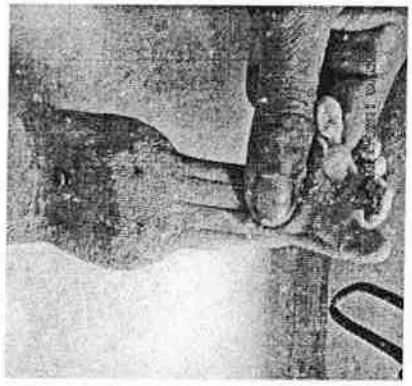

FIG-4