

Sirenomelia within a one-year period at Fallujah general hospital: two case reports

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Abstract: Sirenomelia also called as Mermaid Syndrome, is a rare congenital malformation of uncertain aetiology. It is characterised by fusion of the lower limbs and commonly associated with severe urogenital and gastrointestinal malformation. It should be suspected in antenatal period in cases presenting with severe oligohydramnios and intra-uterine growth retardation. This is a report of 2 cases of Sirenomelia in Fallujah general hospital within a period of about 1 yr., with an incidence of about 10.3 / 100.000 total births which was about 2 to 3 times the registered overall incidence.

Keywords: Sirenomelia, Fallujah General Hospital

1. Introduction

A previous communication reported results of an epidemiological study in Fallujah, western Iraq which revealed very high levels of cancer and infant mortality in the period 2005-2010 together with a statistically significant perturbation of the birth sex-ratio of those born from 2005. The authors concluded that some genotoxic exposure had occurred at the same time as the United States-led attacks on Fallujah in April and November 2004¹.

An attempt to investigate the genetic familial causation of four individual cases of congenital anomaly has subsequently been made², the authors concluded that in these cases there was no familial genetic basis and that sporadic untargated effects were responsible, causes which they described as epigenetic.

It was clearly of interest then to further examine the evidence of increased infant mortality and congenital conditions in Fallujah and to investigate the environment for possible agents that may explain the increased incidence of birth defects and cancer rate increases. The environmental aspect of the problem was studied in 2010 when hair sample analysis of 52 elements was carried out on the parents of children diagnosed with congenital anomalies³. In addition soil and water samples were analyzed. Results indicated that man-made slightly enriched uranium was present in soil and in the hair of the parents, with a trend of increasing concentration along the hair strands back to 2005 but that

no other genotoxic elements were present which could explain the levels of cancer and congenital disease³.

The sirenomelia is a polymalformative syndrome where the most noticeable (but inconstant) aspect is the fusion (more or less complete) of the lower limbs. It is a rare syndrome (1.5 - 4.2 per 100,000 births)⁴ which continues to cause many controversies concerning its etiopathogenesis. Its classification as a variant of caudal regression syndrome (CRS) is still debated⁵. The same applies regarding its relationship with narrow pelvis syndrome and VATER (vertebral defect, anal atresia, interauricular communication; interventricular communication, tracheal and oesophageal atresia, and renal or radial agenesis) syndrome⁶.

The precise aetiology of sirenomelia is not known but many theories have been proposed. Sirenomelia is invariably fatal because of bilateral renal agenesis which leads to severe oligohydramnios and lung hypoplasia. There is no treatment available for sirenomelia and prevention should be the goal. so prenatal diagnosis is very important. It can be diagnosed as early as 13 weeks of pregnancy^(7, 8). Third trimester ultrasonographic diagnosis is impaired by severe oligohydramnios whereas during the second trimester the amount of amniotic fluid is sufficient to allow diagnosis. If diagnosed early, termination of pregnancy can be offered relatively safely to the mother. This would be less traumatic both physically and mentally to the parent than if detected later in pregnancy and moreover because of its association with maternal diabetes mellitus, the maternal blood glucose

levels should be optimal in preconception period and in first trimester to prevent this anomaly.

2. Case Reports

2.1. Case 1

A 2750 gram, term baby born by CS delivery in 3-12-2009 in Fallujah general hospital, to a non-hypertensive, non-diabetic mother living in Al-Resalah district, Fallujah city, aged 31 years & the father is 32 years old, no history of consanguinity between them, & there is no history of such condition in the family. She is gravida 1 para 1, the abnormality have not been diagnosed by prenatal ultrasonography. There was no history of any febrile illness during pregnancy, no history of using teratogenic drug, no smoking and no history of x-ray exposure.

The baby born alive, severely cyanosed & dyspneic. On physical examination; the face, upper limbs & trunk, look apparently normal, there is complete fusion of the lower limbs from the pelvis to the heels with presence of both 5 digits feet which are medially rotated, with absence of external genitalia and anal orifices. Fig 1, 2.



Figure 1. Back.



Figure 2. Front.

The baby died during resuscitation, there was no time to do x-ray or ultrasonography to check for presence of other anomalies. complete fusion of the lower limbs from the pelvis to the heels with presence of both 5 digits feet which are medially rotated, with absence of external genitalia and anal orifices.

2.2. Case 2

A 2500 gm. ,term baby , born by C.S delivery after an uneventful pregnancy to a non-hypertensive non diabetic ,gravida 1 para 1 woman, no history of TORCH infection , no history of using teratogenic drugs or x-ray exposure during pregnancy , she live in Fallujah – Nazzal district

and gave history of regular antenatal visits , with ultrasonography done 3 times in a private clinic in Baghdad & strangely the anomaly was not diagnosed , she is 19 years old & the father is 24 years old , they are consanguineously married ,but there was no family history of sirenomelia or any other congenital anomalies.

The newborn at arrival in neonatal care unit was hypothermic and cyanosed. Clinical evaluation following resuscitation revealed, normal head & face, short neck, normal upper limbs, fused lower segment of the body below pelvis into a single limb with no feet ,Posture of the lower torso was that of alphabetic letter(L)when viewed from the back. There was no opening for urogenital system and anal opening was also absent (figure 3).



Figure 3. Fused lower segment of the body into a single limb with no feet , urogenital & anal openings are absent.

Radiography showed scoliosis of lumbar vertebrae & the pelvis attached to two adhered femurs, no tibia and no fibula (figure 4).



Figure 4. scoliosis of lumbar vertebrae & the pelvis attached to 2 adhered femurs, no tibia and no fibula.

The baby died about one hour after its birth.

3. Discussion

Sirenomelia is a rare congenital anomaly characterized by partial or complete fusion of lower limbs and usually associated with other severe anomalies. It is considered, by many authors, as severe form of caudal regression syndrome⁹. The associated anomalies may include bilateral renal agenesis, complete or partial agenesis of genitourinary system, imperforate anus, absence or ambiguous external genitalia, single umbilical artery, lung hypoplasia and vertebral and cardiac anomalies¹⁰.

In our cases the associated anomalies were complete agenesis of urogenital system and imperforate anus, we couldn't do ultrasonography to check for other abdominal or pelvic anomalies because of the bad condition of the two newborns at presentation.

Etiology of sirenomelia is uncertain and various theories have been proposed to explain its origin. An embryonic insult to caudal mesoderm between 28-32 days of gestation and vascular hypo-perfusion has been proposed as possible factors. Others associated the condition with maternal diabetes mellitus, exposure to teratogens and genetic predisposition¹¹. Sirenomelia carries a grim prognosis, with survival dependent on the nature of the visceral anomalies. As long as there are no severe cardiac or central nervous system defects, lethality is generally secondary to obstructive renal failure due to renal agenesis or dysgenesis¹².

The spectrum of malformation of the lower limbs seen in babies with sirenomelia ranges from fusion of the legs into one lower limb with only two bones present in the entire limb (a femur and a tibia) and absence of foot structures as it is in our 2nd patient to fusion of the skin of the lower limbs along the inner leg with fully formed and separate lower limb bones and fully formed feet which are fused at the ankles¹², this latter form was the case in our 1st patient.

Since the prognosis is bad, the management of sirenomelia is difficult, with unpredictable results. It seems more than justified to put the emphasis on antenatal diagnosis in order to choose the cases with better prognosis. Antenatal diagnosis is based on a morphologic ultrasound study (oligohydramnios, bilateral renal agenesis, a single lower limb, and a unique umbilical artery, absence of a bladder, undetermined external genitalia, anorectal atresia, and lumbosacral agenesis). In sirenomelic foetuses, bilateral renal agenesis causes severe oligohydramnios, hindering a reliable sonographic evaluation of the lower extremity in the second and third trimester. In some cases, bilateral renal agenesis is the only sonographic finding. Amniocentesis and high-frequency transvaginalultrasonographic probes have proven to be very useful in such a situation. Whereas oligohydramnios is a marker of renal agenesis or non-functioning kidneys in the second half of pregnancy, in earlier gestational stages, other contributors to the production of amniotic fluid production are present. Therefore, early in the second trimester, the amount of amniotic fluid should be sufficient to allow detection of sirenomelic abnormalities. Early diagnosis will allow termination of the pregnancy at an early stage, with minor risks and discomfort for the patient¹³.

In our hospital two cases of Sirenomelia were reported at the period between the 1st of December 2009 to the end of November 2010, out of about 7332 live births giving an incidence of about (10.3 / 100.000 births) compared to the overall incidence of 1.5- 4.2/100.000 births⁴. Detailed history was taken from the 2 cases, both gave no history of Diabetes mellitus or any other chronic illness, there was no history of using any teratogenic drugs, & no prior family history of such or any other anomaly in the 2 cases which

means that the occurrence of the 2 cases is sporadic & may be related to environmental factors. History of consanguineous marriage reported only in the 2nd case.

There was failure to diagnose the anomaly by prenatal ultrasonography in the 2 cases because the hospital lacks the ultrasound machine & other facilities needed to for early detection of fatal anomalies.

Accordingly there should be serious plans & steps to be taken by all authorized people & institutions to provide a good antenatal care including good assessment of high risk pregnancies, and to establish a well advanced centre of fetal medicine in Fallujah & provide a good chance for training doctors to offer the right diagnosis of such & other anomalies early in pregnancy.

4. Author's Contributions

This work was carried out in collaboration between all authors; STA designed the study, wrote the protocol & wrote the 1st & final draft of the manuscript & managed the literature searches. AAM examined the 2 cases, offered genetic counselling & shared literature review. Both authors read and approved the final manuscript.

5. Consent

Consent for publication from both sets of parents have been obtained.

6. Ethical Approval

Ethical approval was granted by the scientific committee in Fallujah hospital.

7. Competing Interests

Authors have declared that no competing interest exists.

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