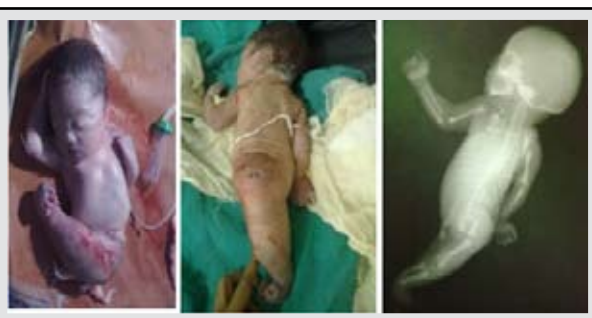
SPOT DIAGNOSIS (IMAGE GALLERY)



BIRTH DEFECT (MERMAID SYNDROME)

Rupa Biswas, Sudip Chatterjee, Milan Ruhidas, Shyamal Banerjee

Department of Pediatric Medicine, NRS Medical College, Kolkata, India.

Address for Correspondence: Dr Sudip Chatterjee, 162, 2 PGH Shah Road,Kolkata-700032. India. Email: sudip_ron@yahoo.com

This neonate was born to a mother addicted to tobacco chewing and also had been on oral contraceptive pills for one year prior to pregnancy. In this child, lower limb is found to be single, malrotated, fused all along the length with 6 toes, flexion at the hip joint is also found. There is absolute absence of any external urogenital orifice but an opening is found posteriorly. Both the upper limbs are found to be normal except radial deviation at the left hand. Potter's facies are present. Infantogram shows fused shaft of femur and absence of both fibulae. On autopsy, a single umbilical artery is found arising from abdominal aorta. The aorta could not be traced below

the origin of umbilical artery. Both kidneys were found in iliac fosse. The kidneys are polycystic macroscopically. Both the kidneys are supplied by branches from abdominal aorta. Both the adrenal glands are hypertrophied. Bladder is rudimentary with 2 ureters opening in the bladder. Small guts are found to be normal but caecum is found to be dilated and ended in a blind loop. The distal portion of the gastrointestinal tract beyond caecum is absent upto the rectum which is found to be atretic. An insignificantly small dimple like structure at the region of anal orifice is present. No reproductive organ found either.

Spot Diagnosis

Mermaid syndrome or sirenomelia. It is an extremely rare congenital disease leading to formation of various internal and external defects in the design of human body. Originally described by Rocheus in 1542 and Palfyn in 1543, this syndrome is characterized by fusion of lower limbs which is also associated with anomalies like flexion, external rotation, symelia and atrophy. Moreover, in this syndrome, there are some lethal defects such as absence of external genitalia, agenesis or any other anomaly of one or both kidneys as well as other gastrointestinal defects. Another pathognomonic finding is the presence of single umbilical, persistent vitelline artery which is the chief distinguishing anatomic finding from Caudal Regression Syndrome. The prevalence of this syndrome is 0.1-0.25:10,000 in normal pregnancies. This syndrome has a strong association with maternal diabetes where relative risk is 1:200-250 and up to 22 percent of fetuses with this anomaly will have diabetic mothers. Some scholars point it to be due to teratogenicity. Animal study confirmed the association with cadmium, retinoic acid, cocaine or irradiation, though convincing data regarding this fact are few. Also modern reproductive procedure such as intracytoplasmic sperm injection was also mentioned as a causal factor.

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