CASE REPORT

Mermaid syndrome: Rare genetic anomaly

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Abstract

Mermaid syndrome (Sirenomelia) is a rare fatal congenital anomaly. The prevalence is reported to be 1 in 100,000 births. The baby appears to have a fish-like tail and joined legs featuring a mermaid at the time of birth or during antenatal screening. Most of these patients die shortly after birth, denoting rare survival rate. Gastrointestinal and genitourinary obstruction with single umbilical artery are the clinical symptoms. Two important hypotheses support the Sirenomelia disorder: the artery steal hypothesis, called the vitelline artery, and the blastogenesis defect hypothesis. There is no known reason for MS, but certain risk factors need to be identified, which include mother aged more than 40 years or less than 20 years at the time of giving birth, cousin marriage, exposure to teratogenic agents, and family history.

A case of this rare congenital disorder was seen in Civil Hospital Faisalabad, Pakistan, which was referred from Duniyapur, District Lodhran, Pakistan. The neonate presented with fused lower limbs, congenital heart disease, and high-grade fever. The mother had a history of gestational diabetes mellitus and hypertension. The baby had fused legs, unidentified internal and external genital system, thumbs anomaly, bile in vomit, and despite life-saving efforts, the patient expired after five days of birth. There is lack of information regarding symptoms and a lack of prenatal screening for MS. Hence, there is a need to create awareness among health care professionals to identify the disease on screening for early diagnosis.

Keywords: Mermaid, Teratogenic, Anomaly, Vitelline artery, Congenital.

DOI: https://doi.org/10.47391/JPMA.6073

Submission completion date: 15-03-2022

Acceptance date: 29-09-2022

Introduction

Sirenomelia or mermaid syndrome (MS) is an uncommon or rare congenital anomaly, in which the neonates have fused legs and perianal area due to mutation in genes and defects in the caudal region of the foetus. This disease was first identified in 1542 and named as such because of the fish-like tail.[1] MS has a rare prevalence rate because of its high mortality and low survival rates. The prevalence of this disease is 1 in 100,000 births.[2] An anomaly scan can identify most of the anomalies during the second or third trimester. Only symptomatic treatment along with major surgeries for leg separation and genitourinary system functioning is available.[3] There is ignorance about the symptoms and a lack of prenatal screening for MS. Hence, there is a need to create awareness among health care professionals to identify the disease on screening for early diagnosis.

Case Report

The case was identified in District Head Quarter Hospital, Faisalabad, in the neonatal ICU on June 12, 2021. The patient was referred from Lodhran Government Hospital by a paediatrician. The neonate was delivered with fused and single lower limb and vague appearance of the genitourinary system from a mother aged 41 with a history of gestational diabetes and gestational hypertension without any anomaly scan in the antenatal period. The neonate was the second child of the family. Before that, the mother had given birth to a normal baby girl, without any anomaly. The family had no history of the said anomaly. The mother had no history of any disease or treatment from any health care centre, and had not accessed any antenatal clinic. The parents lived in a village in district Faisalabad, and the baby was born in a village in Lodhran district. The neonate's weight was 3,000g at the time of birth.

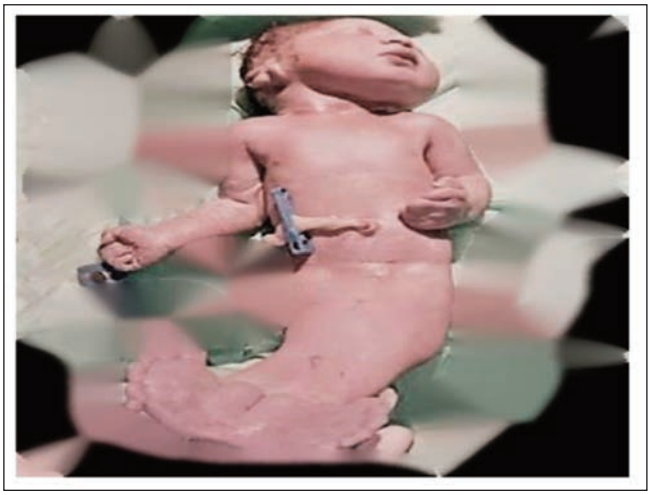

Figure-1: The image shows the appearance of patients with mermaid syndrome. Fusion of fingers, toe, and lower limbs can be observed.

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Table: Neonatal vital signs and blood chemistry in five days.

Days	Pulse	Temp	Oxygen saturation	Blood Pressure	Blood Hb	Blood ESR (erythrocyte sedimentation rate)	Blood CRP
1st	161/m	98F	92% @ 2 litres of oxygen/m	100/60 mmHg	8.8 mg/dl	35 mm/hr	66 mg/L
2nd	164/m	98F	94% @ 2 litres of oxygen/m	100/60 mmHg	8.7 mg/dl	35 mm/hr	68 mg/L
3rd	158/m	100F	92% @ 4 litres of oxygen/m	110/80 mmHg	8.2 mg/dl	55 mm/hr	92 mg/L
4th	130/m	100F	90% @ 2 litres of oxygen/m	110/80 mmHg	7.9 mg/dl	55mm/hr	95 mg/L
5th	180/m	102F	88% @ 5 litres of oxygen/m	Below 70 mmHg	7.9mg/dl	55 mm/hr	104 mg/L

Abbreviations: dl = decilitre, m= minutes, mm= millions per cubic millimetre, L= litres.

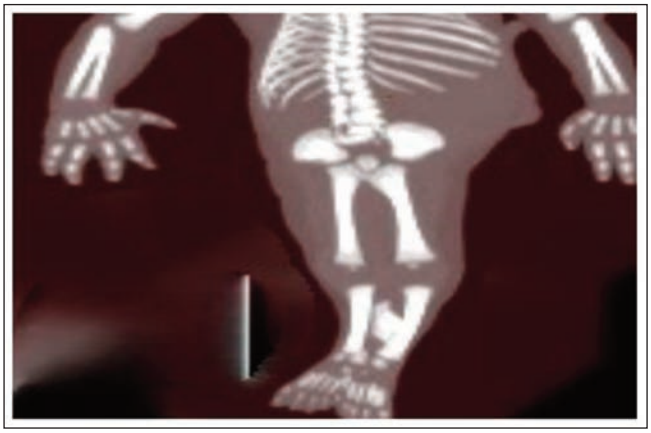

Figure-2: X-ray of patient admitted to the hospital with mermaid syndrome.

The upper part of the body, i.e. the head, face, neck, ears, chest, and abdomen, was normal (Figure-1). On examination the upper limbs were also noted to be normal, but the ring finger of the right hand was smaller than normal. The patient was vitally stable (Table). On auscultation, the heart and lungs sounds were also normal after the birth. After palpitations, the liver was normal in size. No splenomegaly or hepatomegaly was observed at the time of birth.

An artery and vein were present in the umbilical cord. There was a deviation in lower limb fan like appearance of fingers and had 3 heels. There were two thumbs and eight fingers with nails like arrangement. A deep pit-like appearance in the spinal region was noticed at the back of the neonate. Sacral bone prominence was observed in the sacral region, while pelvic bones were fused. A single limb 4cm lower than the anterior cavity was seen locally.

After an echocardiogram and X-rays, the heart was observed to be of normal size and shape. Blood vessels were also normal. A computed tomography scan (CT scan) showed normal liver, bile, while prostatic fluid was also normal. There was a large-sized right kidney and a small-sized left kidney. The urinary bladder was smaller than normal and had an abnormal shape. The internal genitalia testicles or ovaries showed an ambiguous picture. The larger diameter of the umbilical artery had irregular branches. Colour Doppler sonography, showed normal liver, spleen, and bile duct. But the size of the right kidney was smaller than normal and the left was larger than normal. The radiograph showed fused sacral and pelvic bones. There was a fusion of femur bone heads but identified as two bones, and two tibias and fibula with two toes were present. There was a small lower abdominal opening for urinary excretion but had no opening for faecal excretion. The neonate developed abdominal distension, shortly after birth. The neonate had intolerance to oral feed and vomiting, thus parenteral nutrition was given. Due to bowel and urinary obstruction, the patient developed peritonitis. Three days after the birth, the patient started vomiting bile along with faecal contents. The patient expired after the fifth day of birth. While the parents did not agree to have an autopsy done, they agreed to our writing the case report without their names, after explanation.

The patient had reduced blood supply in the lower limbs, had inappropriate nutritional balance (less than body requirement), sacral agenesis, and absence of the urethral opening.

The case was reported in the neonatal ICU, a four-day-old neonate with congenital anomaly, fused lower limbs, ambiguous genital areas, renal abnormality, hyperthermia, and abdominal distension. Treatment attempts included proper oxygenation, control of hyperthermia, administration of antiemetics, blood transfusion, and planning for surgical reconstruction of the lower limb. The patient suffered cardiac arrest and expired despite cardio-pulmonary resuscitation (CPR) on the fifth day after birth.

Discussion

Mermaid syndrome is a rare genetic disorder in which abnormal development of the caudal region result in fusion of lower limbs or fusion of genitalia with presence of bony defects.[4] Studies indicate multiple predisposing factors in the development of mermaid syndrome. Renal failure and hypo-plastic lungs are the causative factors of death. The kidneys in the present case could not be further assessed due to parents' decision to forego autopsy.[5]

Multiple theories have been proposed to describe the aetiology and pathology of mermaid syndrome.[6] Steveson

et al suggested that there is shunting of blood through the abdominal artery which shows abnormality and perforation of the artery at the end of the caudal region. This situation causes missing arteries and veins. The perforation leads to agenesis of caudal structures.[7] The vertebral agenesis leads to dysfunctioning or agenesis of the lower limb. Jaiyessimi et al suggested that sirenomelia cases without artery steal may have other factors associated with the pathogenesis. Teratogenic factors also have a major association in which retinoic acid, cyclophosphamide, and cadmium cause sirenomelia in animals. A rare congenital anomaly, mermaid or sirenomelia syndrome, was first reported by Rocheus et al in 1542. Palin also reported in it 1543. The word Mermaid is associated with a fish-like appearance.[8]

The actual causative factors of mermaid syndrome are unknown, but studies support two hypotheses. According to the Vitelline artery steal hypothesis, the main coeliac artery is short and separated from the aortic arch of the abdomen, and hypoplastic artery of aortic branches or absence of aortic arteries, thus discolouration of a vitelline artery due to loss of blood supply from the caudal part of the embryo and diversion of blood flow from placenta. This process disturbs the third and fourth weeks of ejaculation in the caudal region of the foetus.[9] Bone morphogenic protein stimulate the caudal portion's endothelial cells, production of tissue, vascular system, and normal growth and development in the abdominal region as well as limbs in the embryo.[10]

Most common anomalies in mermaid syndrome include fusion of the lower limbs, defect in omphalocoele of Cantrell, meningomyelocoele, pulmonary hypoplasia, and cleft palate. Mother's age less than 20 or above 40 years, along with gestational diabetes and hypertension are important risk factors.[11] Teratogenicity in pregnancy, such as smoking, environmental hazards, drug abuse, alcoholism, use of tobacco, excessive use of caffeine, and radiography. Symptoms that should be observed in ultrasonography include lower limbs fusion, renal atrophy or agenesis, single umbilical absence or fusion of genitalia, renal insufficiency, and imperforated anus, single lower limb, and foetal heart rate.[12]

In 1961, it was associated with caudal regression syndrome (CRS) due to the similarity of symptoms with CRS anomalies. But, now it is diagnosed as a separate syndrome. Patients with mermaid syndrome have typical appearance of fused lower limbs, larger umbilical artery which is separated from the upper abdominal artery, and sometimes involvement of aortic branches.[11]

Guidera et al reported a case in 1991 with orthopaedic abnormalities and pelvic dysplasia, fused lower abdominal and genital region, and calcanei osseous fusion with abnormal femurs, fibula, tibia, and general abnormalities.[9]

Their patient appeared with a fusion of lower limbs and imperforated anus. On examination, rectal agenesis was diagnosed. They explained the separation of lower limbs and rectal pouch formation through multiple surgeries after 15 days of birth. They also explained the physical activity of a patient with multiple interventions and the patient started crawling and standing at the age of four years.[13] They reported satisfactory nutritional status. In the recently reported case of Faisalabad, the patient's surgeries were planned but he expired after five days of birth. The parents did not agree for any surgical procedure.

Mermaid syndrome is mainly associated with pulmonary hypoplasia or hyperplasia with multiple anomalies such as renal agenesis, cardiac agenesis, and malfunctioning below the lumbar region. More than 70% cases of mermaid syndrome die before the age of five years. Alive patients have a poor prognosis and complicated life problems. In the case presented in THQ hospital, the neonate born with sirenomelia had renal agenesis, a cardiac anomaly with normal heart rate with passage of time went in bradycardia, at birth the neonate had normal body temperature, and sepsis progressed high body temperature and normal blood oxygen saturation.[7]

Conclusion

Mermaid syndrome investigations identified caudal area haemorrhage, gestational diabetes, gestational hypertension, and exposure to environmental risk factors as causative factors. Antenatal screening and follow-up for blood glucose control with environmental risk factors control are recommended. Antenatal screening may detect the anomaly in-vitro, thus early detection and abortion planning could be done. There is a need to educate people about repetitive cousin marriages. There should be antenatal clinics or camps in rural areas. Proper education about health care and screening during pregnancy are necessary.

Acknowledgements: The support and valuable advice of Muhammad Sohail Ashraf is gratefully acknowledged.

Ethical Approval: From the Duniyapur Government Hospital Lodhran.

Disclaimer: None.

Conflict of Interest: None.

Funding Sources: None.

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