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Case Report

Obstetrics and Gynaecology Section

Tetra-Phocomelia: The Seal Limb Deformity - A Case Report

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ABSTRACT

We report a case of term live baby with tetra-phocomelia born to a 35-year-old $G_3P_2L_2A_0$ with history of consanguineous marriage. She was an unbooked case from a tribal community with no previous antenatal visits. At 39 wk of gestation, she was admitted to our hospital with complaint of pain abdomen and on examination was found to be in second stage of labour. She delivered vaginally a term live 2.5 kg female baby with multiple anomalies. There was no history of drug intake, radiation exposure, maternal diabetes or family history of congenital anomalies to support the occurrence of tetra-phocomelia in this baby. The neonate also had multiple facial abnormalities like hypertelorism, microretrognathia and partial cleft palate. Further investigations revealed no abnormalities of internal organs. At present the baby is being followed up at our paediatric department. The case is reported owing to its rarity and term live birth.

Keywords: Consanguinity, Musculoskeletal malformation, Thalidomide

CASE REPORT

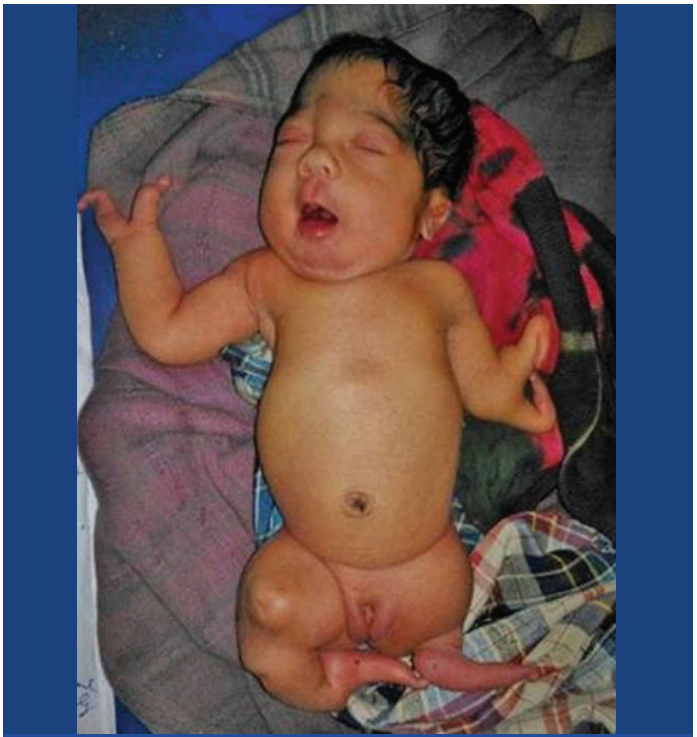
A 35-year-old unbooked $G_3P_2L_2A_0$ at 39 wk 5 d of gestation was admitted to our labour room complaining of labour pain. She had two previous normal vaginal deliveries. There was no history of prior antenatal care and she belonged to a tribal community with low socioeconomic status. She had history of consanguineous marriage and her husband was 40 year old daily labourer. There was no history of drug intake, radiation exposure or fever with rashes in this pregnancy. Neither was she a known case of diabetes nor was there any history of genetic or congenital anomaly in her family.

On examination, she was in second stage of labour with cephalic presentation and regular fetal heart rate. She delivered a term 2500 gm female baby with multiple congenital anomalies [Table/Fig-1]. The Apgar score was 7 and 9 at one and five minutes. Physical examination of the infant showed facial abnormalities like hypertelorism, microretrognathia and partial cleft palate. Right hand appears to be well formed up to forearm with malformed hand and only two fingers while left hand appears to be normal till elbow with malformed forearm and hand having only two fingers. Both the lower limbs are malformed with only one toe in each limb. Examination of placenta and umbilical cord revealed no abnormality. System examination revealed no abnormality. Radiograph of the baby revealed bilateral short humerii with partially formed forearm bones while both the lower limbs showed very scanty attempted formation of femur and tibia with absence of fibula on either side. Ultrasonography of abdomen was normal. Our case is unique as renal anomalies and uterine agenesis were absent in this female baby. Also, there are no associated skull anomalies or pelvic hypoplasia or aplasia in this baby. It is uncommon to see such babies at term at present as antenatal diagnostic modalities help in detecting such anomalies early, offering options of termination. The baby is being followed up by the paediatric and multi-specialty departments.

DISCUSSION

Phocomelia syndrome is a rare congenital malformation affecting the normal growth and development of musculoskeletal system and sometimes can be associated with craniofacial malformation and other system involvement [1]. The term Phocomelia was first coined by Etienne Geoffroy Saint in 1836 and its prevalence is 0.62 in 1,00,000 births [2]. Development of limb bud starts from 26th day of intrauterine life and by the end of 14th wk they are fully formed [3]. Any disturbance in this normal pattern resulted in various types of malformed appendages such as partial (Meromelia) or complete absence (Amelia) of one or more of the extremities [3]. Sometimes long bones are absent and rudimentary hands and feet are attached to the trunk by small irregularly shaped bones called phocomelia (form of meromelia).

Phocomelia can occur as isolated skeletal defect or can be associated with other visceral anomalies like horse shoe kidney, polycystic kidney, cleft palate, hypertelorism and microretrognathia [2]. Based on the various associated clinical patterns of malformation, Phocomelia [4] was further classified in to Al-Awadi/ Raas-Rothschild Syndrome syndrome (AARR syndrome), Roberts/ SC phocomelia, Schinzel phocomelia and Zimmer phocomelia



[Table/Fig-1]: Tetra-Phocomelia "Eye-Hypertelorism, Face-Microretrognathia and Phocomelia in all four limbs"

	Our Patient	AARR* Syndrome	Zimmer Phocomelia	Schinzel Phocomelia	Robert's SC Phocomelia
Limb deficiency	+	+	+/-	+	+
Skull deficiency	-	-	+	+	+
Pelvic Aplasia/hypoplasia	-	+	+	+	-
Uterine agenesis	-	+	-	+/-	-
Cryptorchidism	-(Female)	+/-	+/-	+/-	+
Renal anomaly	-	+	-	+	-
Oral cleft	+	+	+	+/-	+
Abnormal facial features(Including ear, nose and chin)	+	+	+	-	+

*AARR Syndrome: Al-Awadi/Raas-Rothschild Syndrome

[Table/Fig-2]: Different types of phocomelia and clinical features seen in our case

[Table/Fig-2]. Among the various aetiological factors attributed for the cause of this anomaly, thalidomide is a well known teratogenic drug which was prescribed under the label of Contergan and subsequently banned in 1960s [2]. It also affects other systems like cardiovascular, genitourinary, gastrointestinal and nervous system [2]. Antiangiogenic property of thalidomide analogues was suggested as the possible causative factor [5]. Apart from thalidomide, hereditary aetiology like autosomal recessive inheritance was also proposed. Consanguinity was documented in most cases of AARR syndrome [6]. History of consanguinity and the mother being elderly gravida may be the aetiology in our case. Mutations in ESCO2 (establishment of cohesion 1 homolog 2) on 8p21.1 have been reported in Roberts/SC phocomelia [7]. Alp E et al., [4] reported a case of AARR syndrome in a male baby in a nonconsanguenous couple with additional anomalies, such as cryptorchidism, macrophallus and horseshoe kidney. Rao et al.,[1] reported a 20 wk male stillborn fetus with with tetraphocomelia born to a mother with a negative history of consanguinity with additional feature of pelvic hypoplasia. Coodin et al., [8] reported a stillborn male baby with tetraphocomelia, born to a 30-year-old mother at 36 wk of gestation with multiple congenital anomalies like congenital heart disease, absence of extrahepatic biliary tree including gall bladder and common bile duct, polycystic kidneys and megaloureter. Prenatal diagnosis has been reported by as early as 11 wks of gestation in a pregnancy at risk with characteristic, like fusion abnormalities of both upper and lower extremities and a large cystic hygroma over the lower back [9]. Invasive procedures like chorionic villous sampling (CVS) and amniocentesis are useful for prenatal diagnosis in at risk pregnancy cases [9].

CONCLUSION

Our case points to the fact that consanguinity and late pregnancy may predispose to congenital birth defects like phocomelia. Prenatal diagnosis should be offered to all at risk cases and when detected before viability, termination of pregnancy can be offered.

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