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Case Report

TETRA PHOCOMELIA-A CASE REPORT

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ABSTRACT
Tetra Phocomelia is a rare congenital appendicular musculoskeletal malformation with subnormal length of long bones. This phenomenon of shortening is characterized by prominent defect in the proximal part of the affected limb. In severe cases of Phocomelia the hands are direct emergence from the pectoral girdle mimicking flippers of a Seal an aquatic animal a disguise on terrain. This condition was known to be thalidomide syndrome as an attribute to the name of the causing drug during 1960s. It is a tragic toxic effect crusade that queed lakhs of thalidomide babies all over. The present case is of 20 wk male foetus a four limbed phocomelia still born to a 27 year mother with negative history for consanguity, whose musculoskeletal system and other organ systems subjected to evaluate the aetiological factors such as x-ray irradiation, Genetic or sporadic.

Key Words: Tetra Phocomelia, Musculoskeletal Malformation, Thalidomide Syndrome, X-Ray Irradiation and Genetic

INTRODUCTION
Telencephalisation of human brain prompted the evolution of the erect posture which balances the whole body on two hind limbs and also potent manipulating miracle of prehensile upper limbs, the two pivot adaptations delivered all the survival ease benefiting the Homo sapiens to the top of the world. Tetra Phocomelia is a curse depriving the sufferer from the advantage of both the above adaptations completely or partially proportionate to the severity of the case. Phocomelia is a type of Meromelia, in Meromelia there is partial agenesis of limb buds. Tetra Phocomelia is a severe combination of limb defects in which total or partial agenesis of upper and lower limbs is seen, leading to the proximity of limbs to the trunk of the foetus resembling the flippers of a seal “an aquatic animal”. Such babies are termed as Thalidomide babies a consequence of dysmorphogenesis of limb buds due to the use of this drug during 1960. In fact such defects were reported even centuries earlier attributing x-ray irradiation, genetic (autosomal recessive) and sporadic anomalies as causes. Some studies as of Jenna (2009) apply logic and rationale on survival and differentiation theory of progenitor cells defeating the progress zone theory which professed proximodistal patterning (Jenna, 2009) not influenced by the exposure to x-ray irradiation.

CASES
A 27 year female spouse with non consanguinous marriage, G2P1L1A0 with a healthy first female child, with no family history of congenital variations and no history of drug intake during first trimester. The mother gave a history of exposure to x-ray irradiation when she met with an accident. She underwent medical termination of pregnancy in fifth month of gestation as the antenatal obstetric ultrasound scan revealed gross anatomical variation of musculoskeletal system i.e., 4 limbed phocomelia. A 20 weeks stillborn male foetus was obtained from a private nursing home. Postmortem foetogram obtained and foetal autopsy performed. All the features recorded. The foetus weighed about 320grams, with a crown rump length of 16cm (Figure 1).

Features of Axial Skeleton
Normal cranial and facial bones are present, ribs are short and straight when compared to the normal foetus, the vertebral column appeared normal with the growth of the all the components proportionate to the gestational age, the pelvis showed hypoplasia of bones (Figure 2).
Figure 1: Showing the external features of Tetra Phocomelia Foetus

Figure 2: Showing the comparative Radiographic skeletal features of Tetra Phocomelia with a normal foetus of same gestation

The Appendicular Skeleton

Upper limb:
Bilateral short and bowed humeri, forearm bones (radius & ulna)are partially formed which resulted in a typical Phocomelia forearm appearance; both.
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Lower limbs:
Both lower limbs showed short, partially formed femurs, bowing of tibia and absence of fibula on either side (Figure 2).

Other Systems
No abnormalities detected.

Karyotype
Karyotype of the foetus was 46 xy.

DISCUSSION
The drug Thalidomide was banned in 1960. Smithells (1962) reported 15 cases of four limbs Phocomelia with some additional systemic deformities, several cases of tetra Amelia “complete absence of all the four limbs”. Vinod and Shepur (2008) of Karnataka Institute of Medical Sciences reported a case of four limbs Phocomelia. According to Wynbrandt (2003) this deformity occurred due to the dysmorphogenesis of the limb buds during 4th and 5th week of gestation. Mesomelic dysplasia a variation of Phocomelia, characterized by the shortening of the middle portions of the limbs, i.e., “radius, ulna & tibia, fibula” Langer type described by Langer (1967). Charles (2005) defined various degrees of phocomelia. Dignan (1967) mentioned the associations of thrombocytopenia and myeloid leukemoid reactions with phocomelia in attempt to draw common abnormal developmental pathway. Phocomelia an Autosomal recessive trait and occurred in individuals carrying single copy of gene that exhibited dyschondroosteosis. Those having two copies of the genes will have mesomelic dysplasia, the causative gene for this has been discovered as SHOX gene in 1998. The gene is not located on an autosome, but on both X and Y chromosomes termed psuedo autosomal loci (Wynbrandt, 2003). Amongst all the trigger stimuli for this sort of deformity, exposure to x-ray irradiation during first trimester by the mother in the present case is the cause for the Tetra Phocomelia, backed by the progenitor cell survival and differentiation theory (Jenna, 2009).

CONCLUSION
Thalidomide days are gone by, neither the genetic (Autosomal recessive) factor nor the sporadic anomaly which induce phocomelia can be intervened for a better outcome, exposure of the undiagnosed pregnant women to x-ray irradiation would not be an inevitable reality if women who are in active reproductive age and who are planning for pregnancy are educated about the effects of x-ray irradiation on the developing foetus to their understanding by instructing the anganvadi workers and the attending nurses to emphasize on the possible malformations.

REFERENCES