Abstract: Localized malformations of the limbs include absence of bones, extra bones, hypoplastic bones and fusions. Complete absence of a limb is called Amelia, almost complete absence (a mere stub remaining)- phocomelia and partial absence- ectromelia; defects may be transverse or axial. Unilateral phocomelia in a 45 year old female patient, who is otherwise asymptomatic, is reported. The embryonic development of limb is discussed.

Key Words: Phocomelia, Unilateral, humerus

Introduction:
Localized malformations of the limbs include absence of bones, extra bones, hypoplastic bones and fusions. Complete absence of a limb is called Amelia, almost complete absence (a mere stub remaining)- phocomelia and partial absence- ectromelia; defects may be transverse or axial. Two main forms of classification were used widely- morphologic and causal. The currently used classification of congenital anomalies of the upper limb is based on that of swanson modified by the congenital malformation committee of the International Federation of Societies for surgery of the hand (IFSSH) in 1983. A case of longitudinal deficiency of humerus in the left upper limb in a 45-year old woman is reported.

Case Report:
A 45-year old woman presented to the dermatology department with tinea corporis lesions on the front of chest. She was found to have an unusually short left upper limb. The left hand was at the level of elbow of the right upper limb. She was born of a non-consanguineous marriage and had a short left upper limb at birth. There was no history suggestive of maternal illness or drug intake during pregnancy. The patient is married and has two daughters who are normal. She is leading a near normal life and is well adapted to her congenitally short left upper limb.

On examination, she was well built and nourished and was of normal height. Left upper limb was obviously short but there was no muscle wasting. (Fig.1) Hand was normal, both morphologically and functionally. Range of movements (ROM) at the shoulder was full and free.(Figs. 2&3) There was no elbow joint. Radiological evaluation revealed absent humerus with fusion of proximal ends of radius and ulna.(Fig.4) Since she is leading a normal life and is asymptomatic of her defective left upper limb, no treatment was contemplated.

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The embryonic limb buds appear at about the 26th day of gestation; by the 30th day the upper limb has started to differentiate into its three segments (upper arm, forearm and hand). In the lower limb, the same process occurs shortly afterwards. By the end of the 6th week the embryo has acquired a recognizable human form. The upper limbs are fully formed by 12 weeks and the lower limbs by 14 weeks. During this period the muscles and nerves also develop and by the 20th week joint movement is possible.

Most of the malformations involving limb reductions are due to embryonic insults between the 4th and 6th weeks of gestation. Maternal smoking has been found to be associated with an increased incidence of congenital deficiencies. Some are genetically determined and these usually have an autosomal dominant pattern of inheritance. The incidence of congenital deficiencies of upper limb is reported to be 1:9400 live births. Currently used classification of congenital anomalies of the upper limb is based on that of Swanson.

I. Failure of formation of parts (arrest of development)
   A. Transverse arrest (common levels are upper third of forearm, wrist, metacarpal, phalangeal)
   B. Longitudinal arrest (including phocomelia, radial or ulnar club hands, typical cleft hand otherwise referred to as part of the spectrum of synbrachydactyly)

II. Failure of differentiation of parts
   A. Soft tissue involvement
   B. Skeletal involvement
   C. Congenital tumorous conditions (includes radio-ulnar synostosis, synphalangism, camptodactyly, arthrogryphosis, syndactyly)

III. Duplication

IV. Overgrowth

V. Undergrowth (thumb hypoplasia, Madelung deformity)

VI. Congenital constriction band syndrome

VII. Generalized skeletal abnormalities

This classification, although giving some insight into the embryological problem, contains some uncertainties. Others, such as Ogino, have proposed improvements to address some of the uncertainties, but they persist. Such classifications are of little practical value in the everyday management of congenital malformations. In phocomelia, there is a longitudinal arrest of development leading to failure of formation of parts and unilateral involvement is very uncommon. In the present case, there is an arrest of development of humerus in the left upper limb. Phocomelia was more commonly reported in the 1960s due to the teratogenic effects of thalidomide. However, there was no history of maternal illness or ingestion of any drugs during pregnancy by the mother of our patient.

Various prosthetic fittings are available but the
treatment should be started in childhood. However, the reported patient had a near normal life in spite of such a gross deformity. The case is reported for its rarity.

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