An 18 years old male born of non-consanguineous marriage was admitted with enteric fever complicated by hepatitis. During routine physical examination we incidentally found that he had congenital deformity of right hand with absence of index, middle and ring fingers and other two fingers were short (Figs 1, 3). His right chest wall was flattened (skin overlying the ribs without underlying pectoralis muscles) (Fig. 2). Right nipple was slightly smaller and elevated in comparison to left. Latissimus dorsi, supraspinatus, infraspinatus and serratus anterior were not involved. Right biceps brachii muscle was hypoplastic. Right axillary hair was present only on arm-side of armpit. Eye movements were normal. No spinal deformity was found. Lymphnodes were not palpable. Blood picture did not reveal any abnormal cell. Chest x-ray revealed right sided unilateral translucency of lung field (due to absence of pectoralis muscles) (Fig. 5). CT scan of chest did not reveal any lung parenchymal abnormality. The case was diagnosed to have Poland syndrome. No musculoskeletal deformity was reported in his family.

Poland syndrome is a unique pattern of one-sided congenital malformation with involvement of chest muscles, subcutaneous tissues, bones and upper extremity. Absence of sternal head of pectoralis major muscle is the minimal expression. Involvement of adjacent muscles, including pectoralis minor, serratus, latissimus dorsi, external oblique, infraspinatus and supraspinatus muscles also has been described. Skin of the area is hypoplastic with thinned subcutaneous layer. Axillary hair may be absent on the affected side. Ipsilateral nipple is often smaller and higher. Breast is generally hypoplastic in females. Anterior portions of the ribs or costal cartilages may be absent. Anterior lung herniation may be present in severe cases. Scapula may be smaller with winging (Sprengel deformity). Arm, forearm and fingers may be shortened (brachysyndactyly). Short and webbed fingers (cutaneous syndactyly) can be seen on the affected side. Moebius syndrome (congenital bilateral facial paralysis with inability to abduct the eyes) or Klippel-Feil syndrome may be associated with it. Spine and kidney malformations are occasionally reported. Most Poland syndrome cases arise sporadically with a negligible risk of recurrence in same family. Very few familial cases suggest that it may have an autosomal dominant inheritance with incomplete penetration. Incidence of Poland syndrome in India is unknown. In USA, incidence is estimated to range from one in 7,000 to one in 100,000 live births. Vascular developmental anomaly with hypoplasia of subclavian artery during sixth week of gestation has been proposed to be the cause of musculoskeletal malformations. Subclavian artery supply disruption sequence (SASDS) occurs when the medial and forward growth of ribs forces the artery into a U-shaped configuration. Males are more commonly affected than females. Right side is affected more commonly (twice) than left. Reason for these differences are unknown. Mild cases remain undiagnosed until puberty when breast tissue and chest muscle mass development become obvious especially in female. Leukemia and non-Hodgkin lymphoma have been described with Poland syndrome. CT scan and MRI can identify chest wall abnormalities and muscles involvement. Reconstructive surgery is helpful for correcting the chest deformity and breast asymmetry.

References