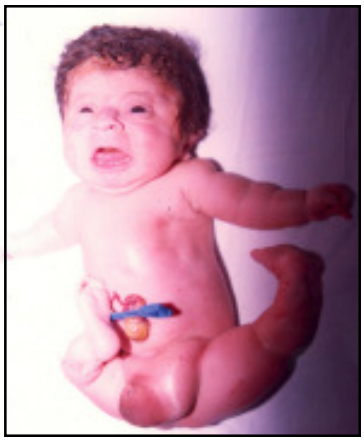

Fig. 1. Full term baby showing micromelia with legs held abducted at right angles to the body.

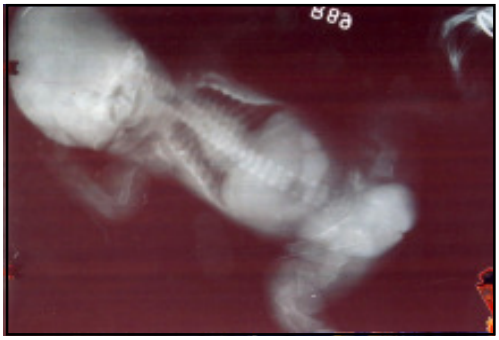

Fig. 2. Radiograph of the baby showing beading of ribs and crumpled long bones of all the four limbs

Discussion

These infants may be still born or die in the first year of life. Birth weight and length are small for gestation age. There is extreme fragility of the skeleton and other connective tissues. There are multiple intrauterine fractures of long bone, which have a crumpled appearance on radiographs. There is striking micromelia and bowing of extremities; the legs are held abducted at right angles to the body in the "frog-leg position". Multiple rib fractures create a beaded appearance and the skull is large for body size with enlarged anterior and posterior fontanelles. Sclerae are dark blue-gray.(2,3,4) The infant we report was 40 minutes old and had blue sclerae, large soft skull with multiple palpable islands, beaked nose, crumpled bilateral femur, multiple fractures of tibia, fibula, humerus, radius and ulna and beaded ribs on radiograph. This baby died at the age of 19 hrs.

The diagnosis is confirmed by collagen biochemical studies using fibroblasts cultured from a skin punch biopsy. Severe osteogenesis imperfecta can be detected prenatally by level II ultrasonography as early as 16 weeks of gestation (1-4).

The morbidity and mortality of osteogenesis imperfecta is due to cardiopulmonary, recurrent pneumonia and transient cardiac failure in childhood and corpulmonale is seen in adults. Neurological complications include basilar invagination but brainstem compression is infrequent. Basilar invagination is best detected with spiral CT of the craniocervical junction. There is no cure for osteogenesis imperfecta. For severe non lethal osteogenesis imperfecta active physical rehabilitation in the early years allows children to attain a higher functional level than does orthopedic management alone. Orthopedic management is aimed at fracture management and correction of deformity to enable function. Fractures should be promptly splinted or cast. Osteogenesis imperfecta fractures heal well and cast removal should be aimed at minimizing immobilization osteoporosis. Correction of deformity of the long bones require an osteotomy procedure and placement of an intramedullary rod (2,3,5).

Treatment with calcium or fluoride supplements or calcitonin does not improve osteogenesis imperfecta.Treatment with biphosphonate drugs is effective in improving mobility and decreasing symptoms in many patients. Intravenous pamidronate or oral alendronate improves quality of life and inhibits bone resorption, thus increasing bone mineralisation, these agents decrease fractures and bone pain (2).

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