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Brittle bones, deformities & deafness

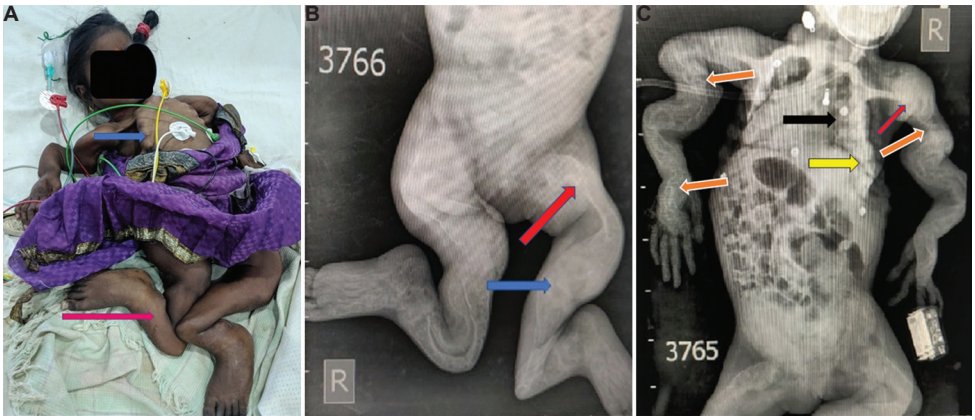

Figure. (**A**) Patient with deformed limbs (pink arrow), beaded ribs (blue arrow) and flail chest. (**B**) X-ray of the lower limbs showing reduced bone density, severe bony deformities with deformed gracile (blue arrow) and protrusion acetabuli with Coxa Vara. (**C**) X-ray of the upper body showing severe kyphoscoliosis (black arrow), multiple fractures (orange arrows), pectus excavatum with broad ribs (yellow arrow) and multiple healing fractures (red arrows).

A 23 yr old female[†], from a remote village Deori, presented to the casualty in Government Medical College, Gondia, India, in March 2019, with severe breathlessness. She had respiratory distress with oxygen saturation of 72 per cent. The patient was fragile and had severe deformities of the limbs, spine and chest (Figure A). She had deafness since birth, normal dentition and white sclera. Skeletal survey revealed severe osteoporosis, multiple fractures, protrusio acetabula with coxa vara deformity and deformed gracile over the tubular bones, typically 'popcorn bones' (Figure B). Chest X-ray showed multiple rib fractures with excessive calluses causing beading (Figure C). She was diagnosed as osteogenesis imperfecta (OI) sporadic type, an overlap between type III and IV with restrictive lung disease. She was put on ventilator and supportive treatment, but she succumbed after three days. Bisphosphonates and gene therapy have been found to improve survival and prevent fractures in OI. This case is presented for its rarity and unique radiological features.

***Conflicts of Interest*:** None.

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[†]Consent to publish clinical information and images obtained from patient's parent.

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