International Journal of Research in Orthopaedics
Gadupudi V et al. Int J Res Orthop. 2023 Jan;9(1):181-185
http://www.ijoro.org

DOI: https://dx.doi.org/10.18203/issn.2455-4510.IntJResOrthop20223010

Case Report

Management of proximal femur varum deformity in skeletal dysplasia: an illustrative case report

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Received: 03 November 2021
Revised: 16 November 2021
Accepted: 18 November 2021

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ABSTRACT

Metaphyseal chondrodysplasia is skeletal dysplasia involving long tubular bones at metaphyseal regions, sparing the epiphysis, predominantly involving lower limbs, hence hindering normal mobilization activity of the individual. Individuals present with pain and deformity due to pathological fracture at the deformity site which necessitates deformity correction and surgical fixation. This is a case of skeletal metaphyseal dysplasia in 47-year female, short statured with bilateral proximal femoral Vara with pathological fracture, treated with deformity correction, surgical fixation. Patient initially presented at 36 years of age with sudden onset left hip pain. She was diagnosed with left femoral neck fracture and coxa Vara deformity. The femoral neck had spontaneously fractured due to increased bending forces acting on the femoral neck due to the coxa Vara deformity. She underwent valgus osteotomy with dynamic hip screw fixation. Further advised for prophylactic corrective osteotomy of contralateral proximal femur however patient was lost to follow-up. At 47 years she presented with pain in the right thigh. She was diagnosed to have right proximal femur pathological fracture due to progressive femur varus deformity for which she underwent right proximal femur valgus closing wedge subtrochanteric osteotomy and fixation using dynamic condylar screw. In our case left proximal femur which was operated did not have any complications. However, right proximal femur which was operated, at four months follow-up revealed no complications except for the foot drop which she developed post-surgery. Hence main aim of the study is to prove that the early prophylactic surgical management of the Vara help to provide successful outcomes.

Keywords: Proximal femur varum deformity, Skeletal dysplasia, Jansen type, Schmid type, Valgus osteotomy, Dynamic hip screw fixation

INTRODUCTION

Metaphyseal chondrodysplasia is skeletal dysplasia involving long tubular bones at metaphyseal regions, sparing the epiphysis, predominantly involving lower limbs, hence hindering normal mobilization activity of the individual. Individuals present with pain and deformity due to pathological fracture at the deformity site which necessitates deformity correction and surgical fixation.[2] This is a case of the skeletal metaphyseal dysplasia in forty-year female, short statured with the bilateral proximal femoral Vara with pathological fracture, treated with deformity correction, surgical fixation. Progressive deformities in the lower extremities are very common in skeletal dysplasia. The pathological fractures occurring as the result of the varus deformities of the proximal femur can be effectively managed by corrective osteotomy and internal fixation. Hence, main aim of the study is to the prove that the early prophylactic surgical management of the Vara help provide successful outcomes.

CASE REPORT

A short statured adult female who presented to our institution in 2008 when she was 36 years old with complaints of left hip pain-sudden in onset, progressive, aggravated with walking without radiation. She did not have any associated trauma. Radiological assessment was done and was found to have left neck of femur fracture (Figure 1 A). Left proximal femur was found to be abnormal with a significant coxa Vara deformity and a mild varus deformity of the middle third of femur. She was taken up for surgical procedure-Left proximal femur valgus osteotomy and dynamic hip screw fixation in 2008 (Figure 1 C). Tissue from the deformity site was sent for histopathological examination which did not show any evidence of tumour/infection. Postoperatively she started on non-weight bearing mobilization and progressive weight bearing as fracture healing occurred. Osteotomy site and the femoral neck fracture healed uneventfully (Figure 2) and patient had returned to her usual activities 6 months after surgery. For the next 12 years she apparently normal without any ambulation difficulties but she presented again in 2020 with complaints of right thigh pain-sudden in onset and progressive without any history of trauma. She did not have any significant birth related complications or delay in motor milestones. Many of her family members including her son were short stature. She did not have any serious health related issues except for hypertension for past 2 years. On examination, she had disproportionate skeletal growth retardation but no mental retardation. Tenderness was elicited over right proximal thigh but with full range of movements of hip and knee. Right lower limb was shortened preoperatively by 3 cm (confined to femur) compared to left lower limb. Her laboratory values showed elevated parathyroid hormone levels with normal calcium and phosphorous levels. Mild derangement was observed in her renal function with elevated serum urea and creatinine. She was diagnosed to have right proximal femur pathological fracture at the subtrochanteric level. There was a coxa Vara deformity (neck shaft angle was 108 deg) and an additional femoral varum deformity of 24 deg at the junction of proximal and middle 3rd of femur (Figure 6 A). Patient required correction of the varus deformity to enable healing of the pathological fracture. It was technically difficult to correct the neck shaft angle and femoral varum as osteotomies at two levels would have been required. Hence it was decided to correct only the femoral varus deformity The preoperative planning is illustrated in Figure 6. We have done right proximal femur valgus closing wedge osteotomy with dynamic condylar screw fixation. Postoperatively there 1 cm lengthening of right lower limb when compared to opposite side. Patient had postoperative foot drop (in peroneal component). Radiological union of the osteotomy site was observed at 4 months follow-up (Figure 10) without any progression of deformity and good functional painless free range of movements of hip and knee achieved. Patient also had good relief of pain. Foot drop was recovering and power had improved up to 3/5 MRC grade at 4 months follow up (Figure 6 D).

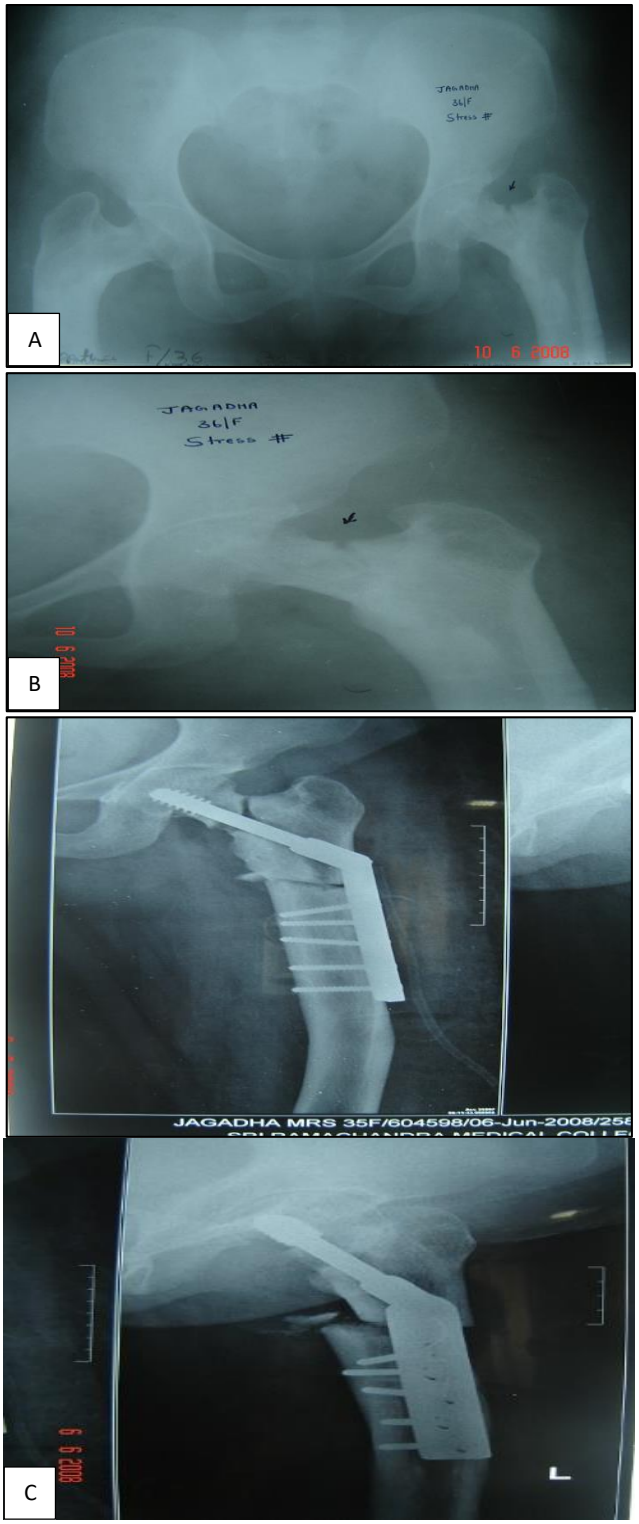

Figure 1 (A-C): X-ray pelvis AP view of 36yr/female with H/O severe left hip pain showing undisplaced left neck of femur fracture in 2008. X-ray left hip AP of 36 year/female with H/o severe left hip pain showing stress fracture on the superior border of neck of femur in 2008. X-ray left hip with proximal femur AP/LAT of 36 year/female with H/o severe left hip pain showing immediate postoperative image showing osteotomy at proximal femur shaft with DHS implant *in situ* in 2008.

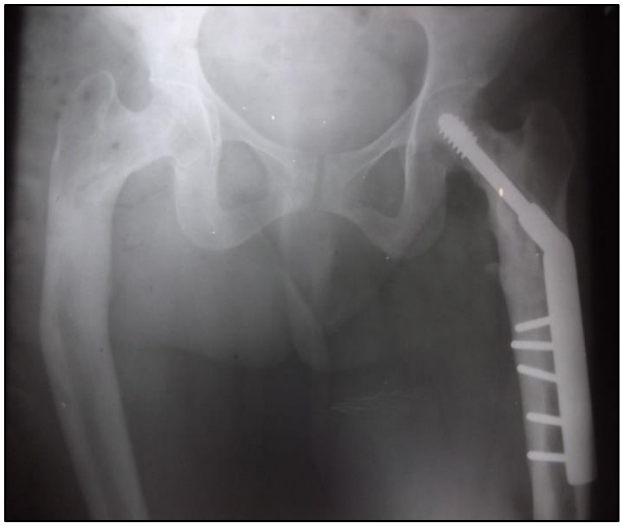

Figure 2: X-ray pelvis with both hips female with H/o severe left hip pain showing coxa Vara deformity and proximal femur varus deformity on right side and left hip DCS implant *in situ* in 2013.

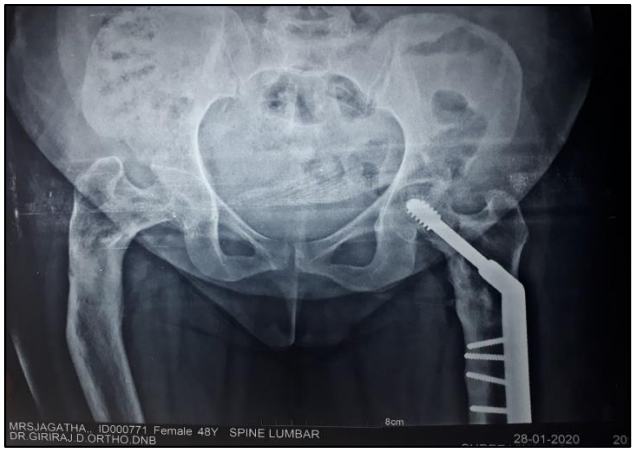

Figure 3: X-ray pelvis with both hips of female with H/o low back pain showing right proximal femur pathological fracture at proximal and middle third junction in 2020.

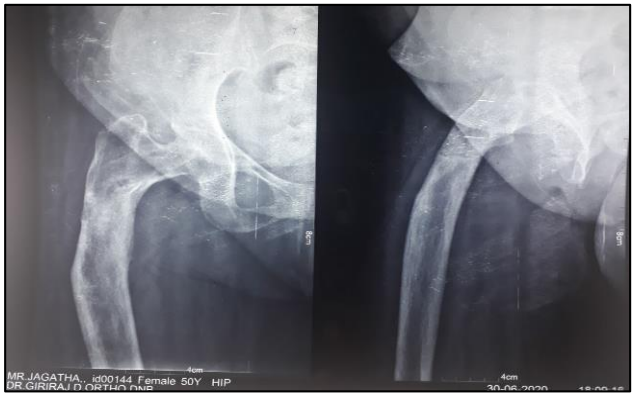

Figure 4: X-ray right proximal femur AP/ lateral of female with H/o progressively increasing severe left upper thigh pain showing dysplastic changes.

Figure 5: X-ray right femur full length AP/ lateral showing coxa Vara deformity with femoral varum deformity at proximal third and middle third junction.

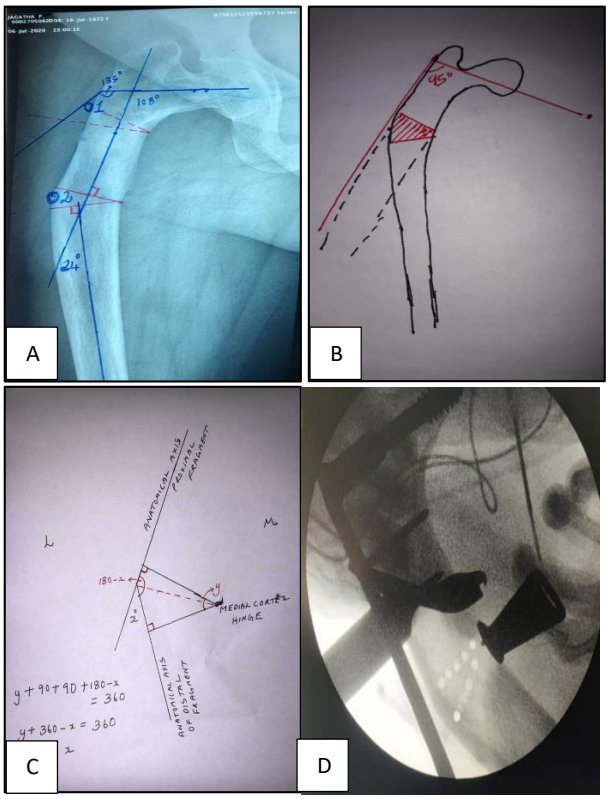

Figure 6 (A-D): Coxa Vara deformity of 108 degree and proximal femur varus deformity of 24 degrees as measured on X-ray. Schematic representation of right proximal femur showing lateral closed wedge osteotomy site. Schematic representation showing angle (y) formed at the medial hinge when femur cuts are made perpendicular to the anatomic axis of proximal and distal fragment is equal to the femur varus deformity (x). Intra operative fluoroscopy image showing DHS implant *in situ* with osteotomy site at the deformity.

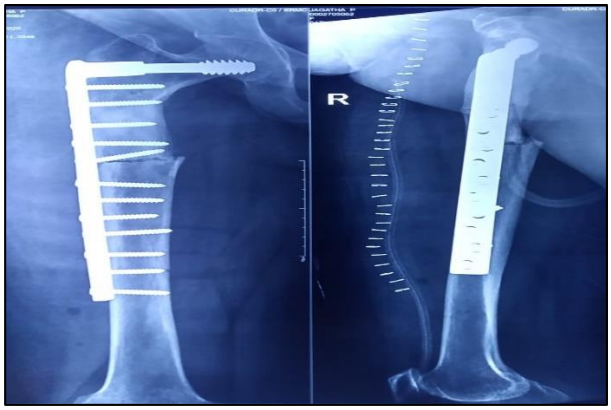

Figure 7: Immediate postoperative X-ray showing DCS implant *in situ* with corrected femur varus deformity.

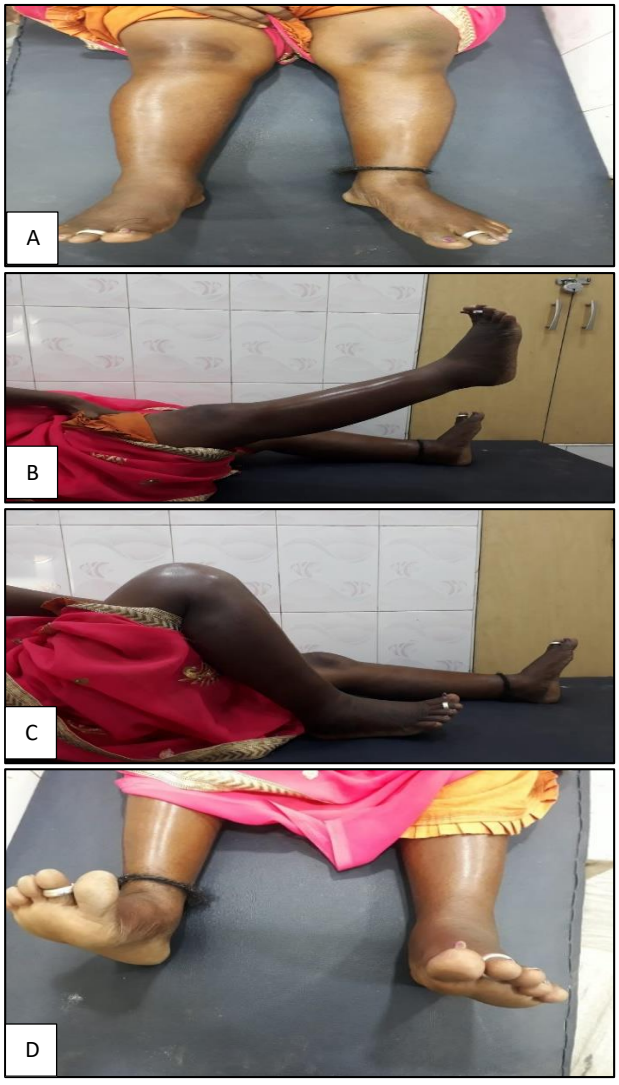

Figure 8 (A-D): Clinical picture after 4 months follow up. 1 cm lengthning of right lower limb compared to left lower limb. No extensor lag or fixed flexed deformity of right knee. Full and free hip and knee flexion movements and right foot drop recovering with MRC grade 3/5 at 4 months followup.

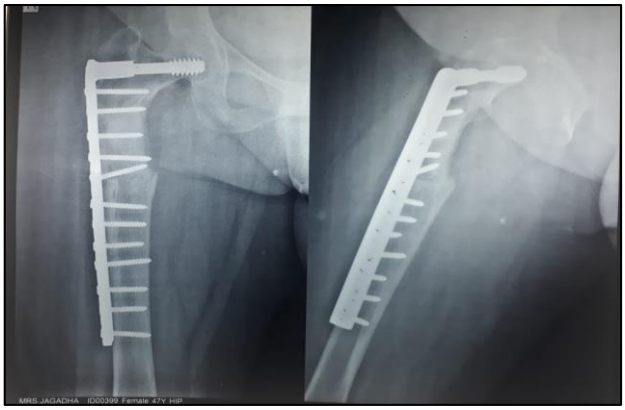

Figure 9: X-ray right proximal femur AP/lateral view showing union at osteotomy site.

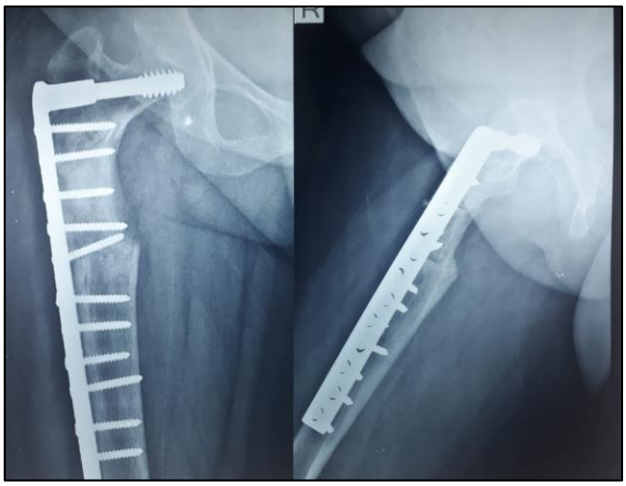

Figure 10: X-ray right proximal femur AP/ lateral view showing dynamic condylar screw implant *in situ* with union at osteotomy site and proper alignment of proximal and distal femur fragments.

DISCUSSION

According to the current available literature, incidence of metaphyseal dysplasia's is about 2 to3 cases per 10000 live births.[1] In our case a middle aged short statured woman who first presented at age of 36 years with stress fracture of left neck of femur was treated with left proximal femur valgus osteotomy and dynamic hip screw fixation. Short statured appearance of this individual along with her extremities being disproportionate with axial skeleton drawn us towards conclusion of skeletal dysplasia. Her laboratory parameters were normal which ruled out any renal related cause like rickets. We also sent intraoperative bone biopsy for histopathological examination which ruled out any infection or tumour etiology. Other clinical features like course facial appearance, waddling gait, exaggerated lumbar lordosis, short arm span, broadened metaphyseal ends of the long bones of forearm, thickened fingers and toes, bowed appearance of bilateral lower limbs suggested diagnosis in favor of skeletal dysplasia. There was involvement of metaphyseal ends of long bones

without any involvement of epiphyseal ends which subclassified our diagnosis into form of metaphyseal dysplasia. She presented 5 years later when progressive varus deformity was observed in right proximal femur for which we advised the need for surgical correction of the varus deformity of right proximal femur, but then she was lost to follow up. She again presented in 2020 when she was diagnosed to have right proximal femur pathological fracture, we have done right proximal femur valgus closed wedge osteotomy with dynamic condylar screw fixation. Now her blood parameters have shown elevated serum calcium levels with mild renal function derangement which coincides with features of Jansen type of skeletal dysplasia.[6] She also developed mild deafness and vision related problems recently. Many of her family members also being short statured which points out that this condition prevails in their family and indicates a genetic correlation exists as in case of skeletal dysplasia's. Genetic testing of the individual nor the family members was done due to logistic reasons.

The general and physical examination of the skeletal dysplasia individuals shows characteristic features like, disproportionate extremities along with deformities, altered gait pattern and facial features, hearing and vision abnormalities.[2,3] These are the key points to identify skeletal dysplastic individual which requires further evaluation to sub classify dysplasia's. Genetic testing is mandatory to identify the correct variant of skeletal dysplasia that helps is providing appropriate treatment. Schmid type dysplasia has defect in type X collagen is a milder form whereas Jansen type dysplasia has defect in the PTH1R receptor is a severe form of disease.[4,5] Hence this helps in assessing the prognosis, morbidity and mortality risks associated with certain form of dysplasia. Metaphyseal dysplasia run in families with either autosomal dominant pattern or autosomal recessive pattern which again signifies the necessity for genetic counselling and screening of the family individuals that helps in early recognition of progressive deformities and appropriate prophylactic surgical fixations that can prevent pathological fractures.[7] Mechanical axis is deviated which is the cause for progression of the deformity. With respect to the anatomical pattern of the proximal femur, more compressive stresses act on the convex side (medial) while tension forces on the concave side (lateral). Hence, varus deformity usually occurs with impending fracture on the lateral aspect which was the scenario in our case. Multi-modality treatment been suggested for individuals with metaphyseal dysplasia's addition to orthopedic surgeon, physiotherapist to guide in strengthening exercises, occupational therapist for mobility advise, pain specialist to avoid activities which strain joints, nutritionist to provide healthy diet and avoid obesity.[2,3]

CONCLUSION

Metaphyseal chondrodysplasia is a form of skeletal dysplasia's which are often progressive and involves the long tubular bone at the metaphyseal regions often sparing the epiphyseal ends. Lower limbs are involved frequently than upper limbs. Pain is the first symptom with which these individuals present for which we must suspect impending pathological fracture.

Progressive deformities in the lower extremities are very common in skeletal dysplasia. Hence surgical fixation of the proximal femur is necessary in case of pathological fracture due to varus deformity.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Gadupudi V, Nalam V, Kadaikaran GHJ. Management of proximal femur varum deformity in skeletal dysplasia: an illustrative case report. Int J Res Orthop 2023;9:181-5.