Correction of Acute Sagittal Plane Angular Deformity of Bilateral Tibia in a Case of Osteogenesis Imperfecta using Limb Reconstruction System

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ABSTRACT

Introduction: Osteogenesis imperfecta is a genetic connective tissue disorder characterized by formation of brittle bones prone to develop fractures and deformities of limbs. Correction of deformity and bone union remains a challenge in such cases.

Case report: Herein we present a case of severe congenital angular deformity of bilateral tibia in anteroposterior plane in a 7 year old girl suffering from osteogenesis imperfecta. She was not able to stand and walk since birth. We treated her successfully by single stage procedure of corrective osteotomy and its fixation with limb reconstruction system (LRS) bilaterally.

Conclusion: External fixation using LRS greatly reduces frequency of fractures and facilitates bone healing effectively in cases of osteogenesis imperfecta. LRS is a excellent tool for correction of deformities in case of osteogenesis imperfecta.

Keywords: Osteogenesis imperfecta, Deformity, Limb reconstruction system

INTRODUCTION

Osteogenesis imperfecta is a group of hereditary connective tissue disorders that mainly affects bones. Term "osteogenesis imperfecta" means the genesis of bones is imperfect leading to formation of fragile bones. Mutations in the *COL1A1* and *COL1A2* genes that encode type I procollagen.[1-3] It is inherited in autonomous dominant fashion and its overall incidence of OI is approximately 1 case for every 20,000 live births. Abnormal synthesis of type I collagen leads to the qualitative defects in bones, teeth, ligaments, sclera and skin. Patients of osteogenesis imperfecta usually presents with history of repeated fractures without significant trauma along with similar kind of family history. There may be considerable differences in severity of expression of various fractures in different patients of the family. We present a case of osteogenesis imperfecta with severe angular deformity in both legs since birth keeping patient bed ridden. We treated this patient successfully by using limb reconstruction system

CASE REPORT

We report a case case of 7 years old girl presented to our department with complain of severe angular deformity of both legs since birth. Patient was never able to stand and walk through out her life. She also had history of repeated fractures in arm and both forearms treated with subsequent plasters. Dentition of the patient was imperfect along with blue sclera. Patient elder brother also had history of repeated fractures in leg and forearms. Patient was having angular deformity in tibia and fibula in coronal plane measuring about 80 degree on right side and 105 degree on left side [Figure 1]. There was no abnormality in central nervous system, cardiovascular system, gastrointestinal tract and respiratory system.Biochemical blood tests were normal except high level of serum alkaline phosphatase. Radiographic analysis of bones revealed healed fractures in both humerus, wrist and anteroposterior angular deformity in mid shaft of tibia fibula, thinning of the long bones and osteopenia [Figure 2]. Diagnosis of osteogenesis imperfect was made on the basis of clinical and radiological findings. Biphosphonate therapy was started and planned for surgical correction of deformities in leg in single stage.

Under general anaesthesia, diaphysis of tibia and fibula was exposed by single anterolateral incision at the site of maximum angulation. Ánterior wedge based osteotomy was performed in tibia at site of angulation along with removal of a segment of fibula at same site. Deformity was corrected slowly on operating table and tibia was stabilised using limb reconstruction system external fixator(LRS). Post operative x ray showed acceptable restoration of longitudinal tibial axis in both limbs [Figure 3]. Clinically patient had almost normal looking well aligned legs at the end of 6 months [Figure 4]. At 6 months follow up x ray of leg showed well united osteotomy site and straight tibia bilaterally**.** LRS was removed at the end of 6 months and patient was followed for

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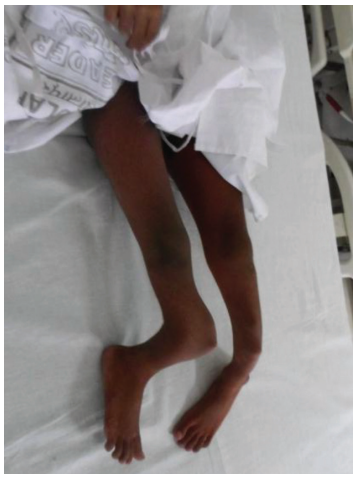

Figure-1: Preoperative clinical picture of both legs showing acute coronal plane angular deformity in right and left leg

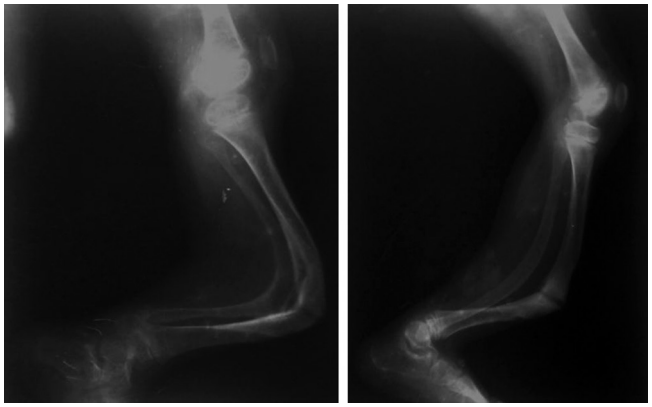

Figure-2: Preoperative X ray of right and left leg-lateral view showing angular deformity in coronal plane.

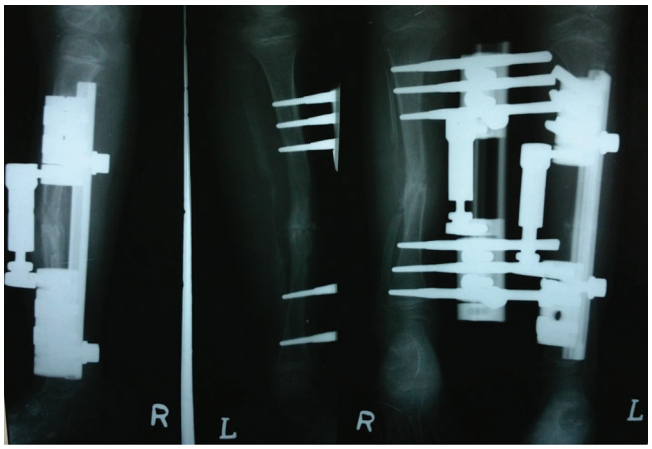

Figure-3: Post operative X ray of both laegs-AP & Lateral views.

2 years. Patient became able to walk without support there was no recurrence of the deformity.

DISCUSSION

Clinical forms of osteogenesi imperfect(OI) vary from mild (OI type I) or moderate (OI type V-VII) to severe features (OI types III and IV), or even lethal in perinatal period (type II). Clinical presentation includes multiple fractures in the absence of significant trauma, multiple bone deformities, blue sclerae, dentinogenesis imperfecta, and conductive or mixed deafness in childhood.[1,4] Quantitative and qualitative defect in type I collagen is attributed to various genetic muatations including *COL1A1* and *COL1A2* genes.[5-8] Most striking clinical feature is tendency to develop fractures in long bone following minor trauma and usually without significant pain and swelling.[6,7] At most instances the clinical presentation and radiographic findings are sufficient for confirmatory disgnosis. However, other diseases responsible for frequent fractures should be excluded. In patients suffering from OI fractures may be discovered during infancy in tends to recur later in childhood but incidence of fractures decreases significantly after adolescence. Although abundant callus formation occurs during healing stage, the quality of this new bone also remains abnormal. This newly formed abnormal bone is more soft and leads to frequent malunion which ultimately results in deformities of long bones. Correction of deformities remains a challenge to orthopaedic surgeons. Loss of reduction, implant failure are frequently noted. Deformity correction in such cases may be achieved by intramedullary nails, ilizarov ring fixator or uniplaner limb reconstruction system. Ilizarov has poor patient compliance as compare to LRS and less technically demanding.

Figure-4: Clinical image of patient at 1 year follow up

CONCLUSION

Osteogenesis imperfecta is a genetic bone disorder leading to genesis of qualitative and quantitative defective type I collagen. Recurrent fractures and abnormal nature of callus leads to formation of deformities of long bones. Correction of deformities by osteotomy and limb reconstruction system is an excellent modality of treatment.

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