Bilateral Genu Valgum due to Chondroectodermal Dysplasia (Ellis-van Creveld Syndrome)

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Abstract

A 14-year-old female with chondroectodermal dysplasia (Ellis-van Creveld syndrome) underwent bilateral high tibial (proximal to the tibial tubercle) osteotomy and deformity correction via circular external fixation for treatment of recurrent genu valgum with accompanying tibial internal rotation and patellofemoral instability.

1 Brief Clinical History

The patient is a 14-year-old female with progressive recurrent bilateral genu valgum (right greater than left), patellofemoral instability, and knee pain (worse in the left) secondary to chondroectodermal dysplasia (Ellis-van Creveld syndrome) with characteristic hypoplasia of the proximal lateral tibial epiphysis (Figs. 1 and 2). Her past medical history included congenital heart disease, which required surgical repair, a lip abnormality, and bilateral complex polysyndactyly of the hands and feet. She had previously undergone two right proximal tibial and fibular osteotomies with acute correction and crossed pin fixation (at age 4 + 0 and 8 + 6) and two left proximal tibial and fibular osteotomies (acute correction and crossed pin fixation at age 8 + 6 and gradual correction with circular external fixation at age 11 + 7). At age 12 + 8, she underwent proximal medial tibial hemiepiphysiodesis via open curettage. She also complains of the knees giving way, leading to falls, related to instability of the patellofemoral joints. She has a mesomelic appearance and is of short stature. Her physical examination reveals full range of motion of the knees, but a positive J sign of the patella.

2 Preoperative Clinical Photos and Radiographs

See Figs. 1 and 2.

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3 Preoperative Problem List

- Bilateral recurrent genu valgum with accompanying tibial internal rotation
- Bilateral incongruity of the knee joint due to proximal lateral tibial hypoplasia
- Bilateral patellofemoral instability

4 Treatment Strategy

Due to complexity of bone deformity bilaterally, our approach was based on sequential limb realignment starting from the more painful left leg. After completion of deformity correction on the left tibia, deformity correction should be performed on the right tibia, while the left tibia is in consolidation. The treatment planning for each limb included a very high proximal tibial osteotomy (above the tubercle) followed by gradual valgus deformity correction and medial translation of the distal tibial segment with the goal of addressing the valgus malalignment of the extremity while simultaneously medializing the tibial tubercle, thereby restoring normal patellofemoral tracking. On both tibiae, the TrueLok circular external fixator was initially applied with two hinges aligned to correct the valgus deformity with medial translation of the distal segment. Completion of the angular deformity correction was followed by frame modification with replacement of hinges and angular

Fig. 1 Front view photograph showing clinical appearance of lower extremities before treatment. Note bilateral genu valgum and accompanying tibial internal rotation
distractor by six hexapod struts on the left tibia to correct internal rotation and by four horizontal translation modules on the right tibia for additional medial translation.

5 Basic Principles

Chondroectodermal dysplasia, also known as Ellis-van Creveld disease, is an autosomal recessive syndrome located at chromosome 4p16.1. The syndrome is typified by polysyndactyly, congenital heart disease, short stature, and lower limb valgus deformity (DaSilva et al. 1980; Ellis and Van Creveld 1940; Fukuda et al. 2012; O’Connor and Collins 2012; Shibata et al. 1999; Weiner et al. 2013). Patients have sparse thin hair, atrophic to absent fingernails and toenails, and abnormal small teeth, all of which are characteristics of ectodermal involvement. The valgus deformity seen in patients with this condition originates in the underdevelopment of the lateral proximal tibial epiphysis. Over time, the distal femur also becomes deformed. Proximal tibial osteotomy in young children is successful in realigning the limb temporarily, but the valgus deformity assuredly recurs due to abnormal proximal tibial physeal growth. Hemiepiphysiodesis of the proximal medial tibia can arrest the progression of the valgus but, due to the overlying short stature of the patients, is not favored in the young child. Tibial torsion is also often present as a component of the deformity.

Fig. 2 Standing AP radiograph of lower extremities demonstrating bilateral complex deformities of the femur and tibia with right genu valgum 22° (LDFA=80°, MPTA=102°, JLCA=0°) and left genu valgum 18° (LDFA=80°, MPTA=112°, JLCA=0°, proximal metaphyseal varus 25°) with hypoplasia of proximal lateral tibial epiphysis on both sides.
and should be corrected accordingly. Over time, the patients develop patellofemoral instability due to lateralization of the quadriceps mechanism and tibial tubercle and become symptomatic in adolescence.

6 Images During Treatment

See Figs. 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, and 14.

7 Technical Pearls

Obtaining a sufficient fixation of the proximal segment after high tibial osteotomy above the tibial tubercle is challenging due to small length of the bone segment and location of the osteotomy proximal to the insertion of the patellar tendon. Application of standard horizontal wire and two (anterolateral to posteromedial and anteromedial to posterolateral) half pins parallel to the surface of the ring is often impossible due to bone size limitations. Placement of wire and half pins obliquely
Fig. 4 AP radiograph of the left tibia after completion of angular deformity correction and medial translation. Note significant internal rotation of the tibia required gradual correction.

Fig. 5 Front view photograph showing clinical appearance of the left lower extremity during rotational deformity correction. Note that hinges and angular distractor were replaced with six hexapod struts (Polihex Hexapod System Litos, Hamburg, Germany) followed by their gradual adjustment according to software prescription.
relative to the ring surface allows maintaining the same stability of fixation while occupying much less of the space (Fig. 3). Due to location of the deformity apex and specific shape of the proximal tibial metaphysis, angular deformity correction should often be combined with significant medial translation. Therefore, when angular deformity correction in those cases is achieved by using uniplanar hinges, the axis of those hinges should be translated far proximal requiring increase rigidity of the connection rods to prevent bending. Sometimes, limb realignment required correction of associated rotational deformities or additional horizontal translation of bone segments necessitating modification of the Ilizarov-type frames during the treatment. The use of hexapod-type circular external fixators allows avoiding frame modification during deformity correction and requires only recalculation of the strut adjustment prescription.

8 Outcome Clinical Photos and Radiographs

See Figs. 15, 16, and 17.
Fig. 7  Front view photograph showing clinical appearance of the left lower extremity during consolidation period. Note that six hexapod struts were replaced by four threaded rods with universal lockable hinges to increase stability of fixation. During the consolidation period on the left tibia, patient was returned to operating room for tibial osteotomy and frame application on the right tibia.

Fig. 8  AP and LAT radiographs of the left tibia at the end of consolidation period.
Fig. 9  AP and LAT radiographs of the left tibia after frame removal. Note improved limb alignment and remodeling of bone regenerate.

Fig. 10 Postoperative AP radiograph of the right tibia after high proximal tibial osteotomy (above the tubercle) and application of the TrueLok circular external fixator. Note that frame configuration and pattern of bone segment stabilization were similar to that on the left tibia.
9 Avoiding and Managing Problems

While the goals of treatment were achieved in this teen, the underlying incongruity of the joint cannot be addressed. Accompanying chondromalacia of the lateral distal femur and proximal tibia was seen on subsequent arthroscopy performed for complaints of knee pain at age 18. There was also an interesting finding on MRI and subsequent arthroscopy of bilateral discoid lateral menisci, which had significant tears requiring debridement. The ultimate outcome of this patient and others with this condition is likely joint arthroplasty in adulthood.

Fig. 11 AP radiograph of the right tibia after completion of angular deformity correction and medial translation. Note that additional medial translation is required for realignment of the tibia

Fig. 12 Photograph of the right tibia illustrating frame modification. Note that proximal ring and distal double-ring block are interconnected by four horizontal translation modules for additional gradual medial translation
Fig. 13  AP and LAT radiographs of the right tibia during consolidation period. Note that horizontal translation modules were replaced by four threaded rods with universal lockable hinges to increase stability of fixation.

Fig. 14  AP and LAT radiographs of the right tibia after frame removal. Note improved limb alignment and remodeling of bone regenerate.
**Fig. 15** Standing AP radiograph of the lower extremities 1 year after frame removal demonstrating restored alignment with complete remodeling of both distraction regenerates

**Fig. 16** Front and side view photographs showing clinical appearance of lower extremities after treatment
10 Cross-References

- Correction of Bilateral Genu Varum for a High Level Athlete
- Spondyloepiphyseal Dysplasia Treated by bi-lateral Proximal Tibial Osteotomy Followed by Gradual Deformity Correction

References and Suggested Reading

Ellis RWB, Van Creveld S (1940) Syndrome characterized by ectodermal dysplasia, polydactyly, chondrodysplasia and congenital morbus cordis: report of three cases. Arch Dis Child 15:65–84

Fig. 17 Standing AP radiograph of the lower extremities 4 years after frame removal demonstrating stable result without recurrence of the deformities