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Case Reports

Correction of knee deformity in patients with Ellis–van Creveld syndrome: A case report and review of the literature

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ABSTRACT

Ellis–van Creveld Syndrome (EVC) is a rare autosomal recessive disorder. In 1940 Richard W. Ellis and Simon van Creveld first reported on a rare skeletal dysplasia, which to emphasize the main clinical characteristics, was termed "chondro-ectodermal dysplasia". The ectodermal involvement includes the skin, hair and nails while the chondrodysplastic characteristics involve the cartilage and bones, primarily in the forearms and lower legs. For the orthopaedic surgeon progressive valgus knee deformity accompanied by patella dislocation is the main problem in EVC. This study reports a ten year follow-up after a primarily failed operative therapy of knee deformity due to incomplete correction and the surgical technique utilized to correct the residual external torsional deformity and dislocation of the patella in a 19 year old girl who presented with the typical clinical features of Ellis–van Creveld Syndrome.

1. Introduction

Ellis and van Creveld first reported in 1940 an autosomal recessive syndrome described as a chondroectodermal dysplasia (CED) [1] later referred to as the Ellis van Creveld syndrome. This is an extremely rare genetic disorder, caused by mutations in two non-homologous genes (EVC and EVC2) in a head to head configuration located on chromosome 4p16 [2]. Globally approximately 50 cases have been described [3]. Usually it is diagnosed prenatally during ultrasonic testing or at birth through the observation of typical characteristics in the skeletal survey of short-limbed dwarfism and post-axial polydactyly or syndactyly [4,5]. Confirmation of the diagnosis of EVC is accomplished by molecular testing.

The relevant gene was first identified in 2000 [6–8]. Radiological diagnostic characteristics include short segments of tubular bones, accompanied by proximal to distal cortical thickness. Chondrodystrophy commonly occurs in the bones of the forearms and lower extremities [7,8]. Clinically, patients with EVC present an above average incidence of polydactyly, short broad hands and feet with short, thick fingers and toes and progressive genu valgum [9]. For the orthopaedic surgeon progressive valgus knee deformity accompanied by patella dislocation and external–torsional deformity of the tibia are the main problems in EVC. In the literature there are only a few and inconsistent reports about operative correction of this severe deformity. There is still no consensus concerning the most effective operative strategy, although it has been found that incomplete correction of the deformity results in recurrence of the deformity [4,10,13].

We present a ten year follow-up and the complex operative treatment in a 19-year-old Turkish girl, with a previously diagnosed and treated EVC, who was referred to our department with increasing impairment of her walking distance and pain due to a progressive medial instability secondary to recurrent dislocation of the patella following a high tibial varising osteotomy performed 9 years ago.

2. Case report

At the age of ten years the patient had undergone a open-wedge varising osteotomy with an Ilizarov fixator to correct progressive valgus deformity of the right knee. Initially there was clinical improvement of her gait and she was able to walk without splints for two years. Due to increasing knee instability caused by developing dislocation of the patella a custom made splint was fabricated and used to maintain patellar stability. The most recent major problem the patient experienced was a gait disturbance and increasing knee pain with substantial medial knee instability. Clinically the right knee showed an obvious medial laxity (Fig. 1) with dislocation of the patella. (Fig. 2). Additionally, the right tibia was externally rotated 70°. The femoral- foot angle was 80°. Mobility of the right knee was 100°/0°/0° for flexion and extension. Radiography of the right knee including a 3-D CT scan of the knee, demonstrated a valgus deformity of the tibial plateau, dislocation of the patella and external-rotation deformity of the lower leg (Fig. 3).

Our aim of surgical treatment was to correct all deformities with a single procedure including the torsional malalignement of the tibia,

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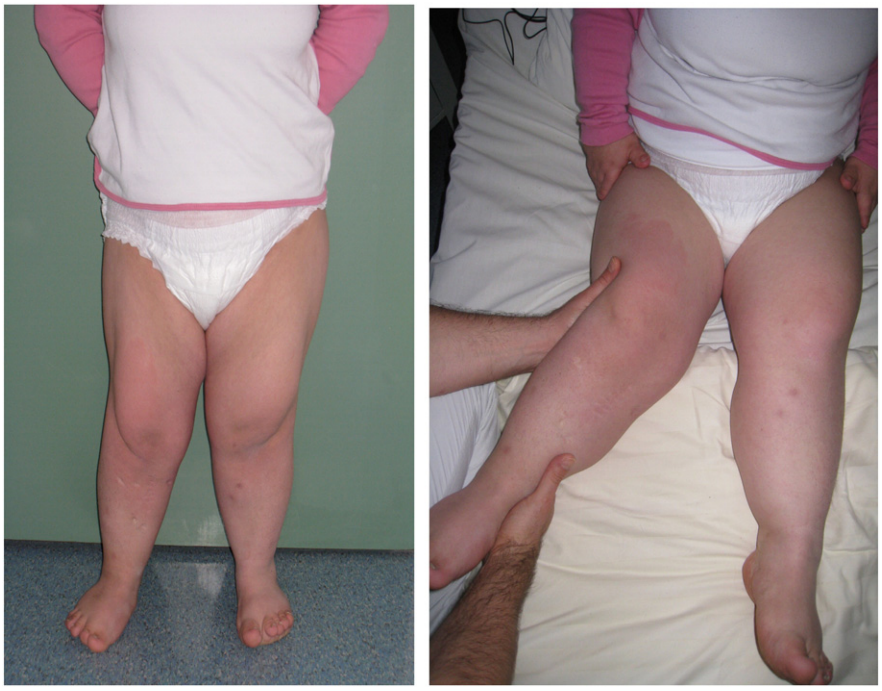

Fig. 1. Preoperative valgus deformity of the right knee due to clinical evident medial instability.

dislocation of the patella, lateral soft-tissue contracture and medial knee instability.

3. Surgical treatment

The surgical procedure, with the patient in the supine position under general anesthesia, was divided into three steps. Initially an antero-lateral approach to the knee was implemented, starting beyond the lateral femoral condyle down to the tibial metaphysis. The first step involved a soft-tissue release of the lateral knee capsule and the lateral retinaculum. The tensor fascia lata was released and a fasciotomy of the biceps tendon was performed to obtain reduction of the patella. In a second step a medialisation of the tibial tuberosity (Elmslie procedure) with reefing of the medial retinaculum was carried out. Finally a 40° tibio-fibular medial derotation osteotomy was performed. Due to the small size an angular stable locking device, 6 hole T-plate, (Synthes, Switzerland), was utilized for fixation.

Because of the medialisation of the tibial tuberosity it was possible to place the T-plate anteriorly. The plate was shaped manually to obtain a good contact face. Temporarily the plate was fixed with 1.6 mm K-wires. Plate position and the alignment after the osteotomy were checked with an image intensifier in both planes. Finally the plate was fixed permanently.

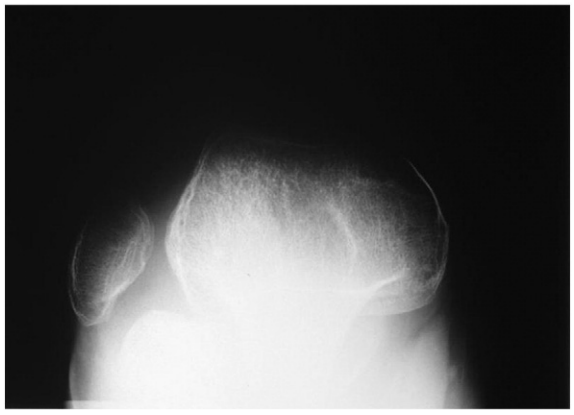

Fig. 2. Preoperative skyline view of right knee shows complete lateral dislocation of the patella.

Postoperatively the right leg was immobilized in a long-leg plaster cast for the first 6 weeks. Wound healing was uneventful. Gentle exercising started from the second postoperative day with passive assisted mobilization at the hip and knee-joint. Active flexion after one week was possible up to 45°. Clinical testing and postoperative radiological imaging one year postoperatively show a centered patella with normal mechanical axis and correction of the external–torsional deformity (Fig.4).

4. Outcome

No weight bearing was permitted for the first 12 weeks. Bony consolidation was first apparent after 6 weeks. At the full leg standing merchant view the Mikulicz-line proceeded physiologically in the joint center. During the outpatient care domiciliary physical therapy was prescribed. On follow-up the knee joint was stable in each position. Passive and active range of motion was 120°/0°/0° for flexion/extension. In comparison to the preoperative range of motion there was no restriction. Postoperatively our patient was able to walk without crutches and to perform one-leg stance after 3 months (Fig. 5).

5. Discussion

Progressive valgus knee deformity is the main orthopaedic problem in EVC. In EVC the plateau of the tibia is poorly developed, typically with a lateral depression [10,11]. This leads to progressive valgus malalignment often combined with medial instability of the knee and lateral dislocation of the patella. In literature there are only few and inconsistent reports, thus operative correction of knee deformity in EVC is still a controversial issue. Milgram et al. reported that the typical valgus deformity recurred after osteotomy of both tibiae and fibulae in a 9 year old patient with EVC [12]. On the first occasion postoperatively, the osteotomies required long leg casting for 2 months, which led to complete union. After reoccurrence of the