Tibial Hemimelia

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Introduction

The first case of tibial hemimelia was reported in 1841 by Otto [1] in the German literature. However, he incorrectly described it as fibular hemimelia, and Burckhardt later recognized his mistake in 1880 [2, 3]. Thus, the first correct reported complete case of tibial hemimelia is credited to Billroth in 1861 [4, 5].

Tibial hemimelia often appears as a shortened leg with knee and ankle deformity. Most often there is a knee flexion contracture, though multidirectional instability may also be present due to a lack of collateral ligaments. A patella and quadriceps mechanism may be present or absent and may have limited function if present. There may be cutaneous manifestations such as dimples over the head of the fibula or the free end of the tibia. The tibia may be shortened, dysplastic, or absent; it may also present as an anlage invisible to radiographs. The fibula can be normal or dysplastic, and often subluxated or dislocated proximally and/or distally. The ankle is often in varus and equinus, and the foot may be adducted and supinated. The medial side of the foot may be missing rays.

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Genetics

The incidence of tibial hemimelia is reported to be one in one million [6, 7]. In 1941, there were 79 published cases [8]; since then there have been several hundred more reported. Tibial hemimelia may be diagnosed on prenatal ultrasound as early as 16 weeks of gestation [9]. There have been many studies on the genetic inheritance of tibial hemimelia. Multiple reports describe parent to child transmission (Nutt [8], Jones [47]) as well as families with multiple siblings affected (Aitken [10], Emami [11]). Autosomal dominant heritance has been implicated by Clark [12], Cowell [13], and Lenz [14, 15]. Autosomal recessive inheritance has also been described (Fried [16], Mahjlondji [17], and McKay [18]). An interesting breeding trial of Galloway cattle with tibial hemimelia concluded that tibial hemimelia was caused by homozygosity of a single autosomal recessive gene with variable expressivity and pleiotropic effects on various body systems [19].

Tibial hemimelia has been found to be associated with several syndromes. Werner's syndrome, or tibial hemimelia–polysyndactyly–triphalangeal thumb syndrome [20] (THPTTS) is an autosomal dominant disorder which is now thought to be a variant of triphalangeal thumb-polysyndactyly syndrome (TPTPS), which have both been mapped to chromosome 7q [21]. It has also been proposed that deletion on chromosome 8q, contiguous with Langer-Giedion syndrome, or type II tricho–rhino–phalangeal syndrome (TRPS II) may also be responsible for tibial hemimelia [22]. Case reports have also linked tibial hemimelia and other limb anomalies to CHARGE syndrome, a pattern of congenital anomalies including eye, nose, ear, heart, and genital defects [23, 24]. Most cases of CHARGE syndrome are due to a mutation of the CHD7 gene (chromodomain helicase DNA-binding protein 7), located on chromosome 8q, and CHD7 is known to be expressed by the developing limb bud mesenchyme [25, 26]. In addition, tibial hemimelia has been linked to tibial hemimelia-diplopodia syndrome [27], tibial hemimelia–split

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hand and foot syndrome [28], and tibial hemimelia–micromelia–trigonal brachycephaly syndrome [29].

Tibial hemimelia can be unilateral or bilateral. It is estimated that 30 % of cases are bilateral [30]. It does seem to favor the right side for unknown reasons. Spiegel noted that all 11 of his unilateral cases affected the right side, and in his review, about 72 % of unilateral cases in the literature affected the right side [31]. Tibial hemimelia has also been associated with multiple congenital anomalies. In Schoenecker's case series of 57 patients, 34 (60 %) had associated anomalies [32]. Similarly, Launois and Kuss found 24/41 (59 %) cases had associated anomalies [33].

In the ipsilateral limb, additional deformities may be suppressive or duplicated. Reported anomalies include congenital femoral deficiency, bifid femur, missing patella or quadriceps mechanism, knee hyperextension or flexion, clubfeet, missing toes, syndactyly, supernumerary or duplicated great toe, and a "mirror foot" [34–38]. Other reported limb deformities include radial dysplasia, lobster claw deformity, hand syndactyly, polydactyly, triphalangism, missing fingers or toes, hip dysplasia, hip dislocation, coxa valga, hemivertebrae, and myelomeningocele [39–42]. Associated deformities that have been reported include deafness, cleft palate, pseudohermaphroditism, cryptorchidism, and hypospadias [30].

Pathoanatomy

Evans and Smith [43], in an anatomic examination of the leg, found both an absence and duplication of some muscles. He described some muscles as being essentially functionless and attached to only one bone. He postulated a mesoblast origin and not likely a mechanical or traumatic source, confirming what Hovelacque and Noel had found in mouse embryos in 1909 [44].

Turker et al. [45] dissected five Jones type Ia specimens. They found ipsilateral toe anomalies in most cases, ranging from four to eight digits. The affected leg had smaller girth, and a dimple was consistently found where the skin was tethered over fibula. They found intact saphenous and lesser saphenous veins, as well as sural and superficial peroneal nerves in the subcutaneous tissues. The deep peroneal and posterior tibial arteries were found associated with their respective nerves, but the posterior tibial bundle was found to be short and acting as a tether. Lateral and superficial posterior compartment muscles were generally intact with normal insertions. Gastrocnemius and soleus muscles had confluent Achilles tendon, which inserted on the medial side of the calcaneal tubercle. The anterior and deep posterior compartments did not have a discrete boundary. Tendons had anomalous courses and sometimes split. No identifiable posterior tibial or anterior tibial muscle belly was found, but all specimens had a tendon inserting medially on the midfoot that tethered the foot in supination. Three specimens had an anomalous tendon inserting onto the neck of the talus. In addition, all specimens had a flat cable of tendon-like structures on the anterior border of the fibula with minimal muscle associated proximally. This unknown structure wrapped around and inserted on the posterior capsule of the ankle. Abductor hallucis muscles were found in all specimens, even in feet without medial rays. No discrete plantar fascia was found. All specimens had subtalar coalitions, and some had midfoot coalitions. The talus articulated with the distal medial fibula on its posterolateral side with a vertical and sagittal orientation with only one plane of rotation.

Box 24.1. Introduction to Tibial Hemimelia

- Incidence is approximately one in one million.
- Autosomal dominant and recessive inheritance has been reported, with variable expression.
- Defects on chromosome 7q and 8q have been implicated, as well as the CHD7 gene.
- Common associated deformities: congenital femoral deficiency, bifid femur, absent patella, mirrored foot, and polydactyly.
- A dimple is often seen on the proximal fibula.
- Anterior and deep posterior compartments of the leg are most deficient, with tendon remnants still attached to the foot.
- The talus tends to follow the fibula in the deformity.

Classification

Frantz and O'Rahilly described a classification system for congenital skeletal limb deficiencies in 1961 [46]. The authors described amelia, hemimelia, phocomelia, and other limb deficiencies. Hemimelia was further described as being complete, partial, or paraxial. They are then divided into terminal (distal) and intercalary (middle) deficiencies, and further subdivided into transverse and longitudinal deficiencies.

The Jones classification (Fig. 24.1) was proposed in 1978 and was based on radiographic criteria. Type I deficiencies had no visible tibia and were subdivided into two groups: the Ia group had a hypoplastic distal femoral epiphysis and the Ib group had normal ossification of the distal femoral epiphysis, suggesting the presence of a proximal tibial epiphysis. Type II deficiencies had a visible proximal tibia but had a distal tibia deficiency. Type III deficiencies had a visible distal tibia, but had a proximal tibia deficiency. Type IV deficiencies had a shortened tibia with significant distal tibiofibular diastasis [47].

Kalamchi and Dawe, in their study of 21 cases in 1985, proposed a modification to the Jones classification and the

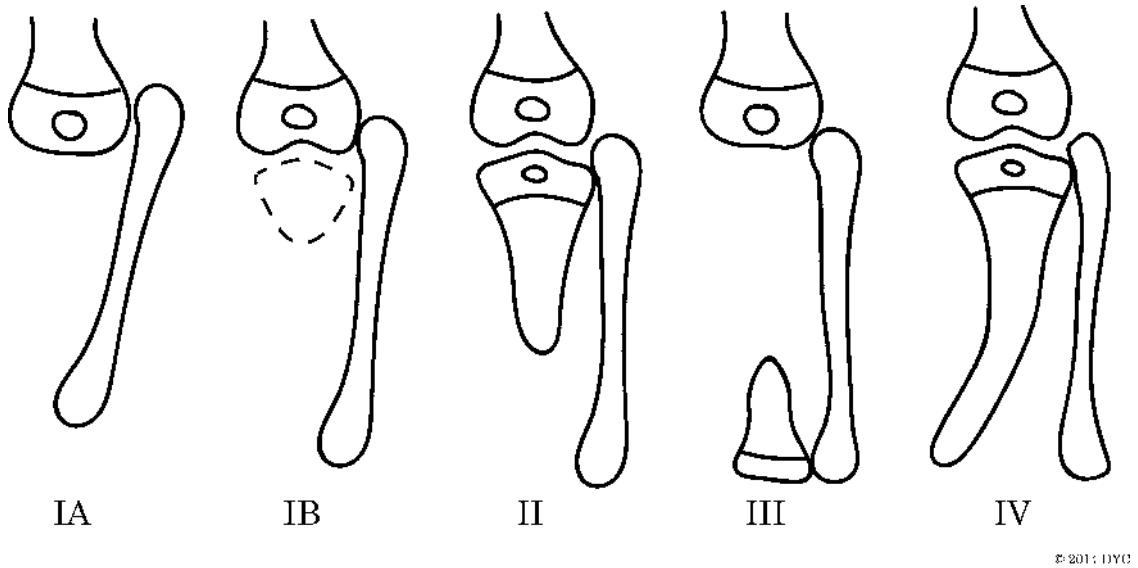

Fig. 24.1 Jones classification of tibial hemimelia

results of their treatments for each type [48]. Type I deficiencies were defined as a total absence of the tibia. This group had knee flexion contractures, greater than 45° and no active quadriceps function. These patients also had proximal migration of the fibular head and hypoplasia of the distal femur. Type II deficiencies were defined as distal tibial aplasia, with a proximal tibia present. There was active quadriceps function and had knee flexion contractures between 25° and 45°. They had more normal development of the distal femur, with normal metaphyseal width and normal epiphyseal ossification, indicating at least a proximal tibial anlage was present. There was also less proximal migration of the fibula. Type III deficiencies were defined as distal tibia aplasia with diastasis of the tibiofibular syndesmosis. They had normal knee joints and good quadriceps function. Usually the talus was subluxated proximally with a prominent distal fibula.

Paley proposed a classification in 2003 based on the Jones classification [49]. The Paley classification (Fig. 24.2) is modified in this manuscript and described in more detail below. The classification is oriented around the treatment for each type.

The most recent classification scheme was published by Weber in 2007 [50]. Previous classifications had not taken into account the cartilaginous anlage. He also added a few types of tibial hemimelia which were previously unclassifiable by the previous schemes and previously published by case report [51]. Weber's new classification took into account the entire affected leg, from hip to foot. He proposed seven types of increasing severity, with subgroups based on whether the cartilage anlage was present (a) or not (b). Type 1 was tibial hypoplasia but intact proximally and distally. Type 2 was distal diastasis. Type 3 was distal aplasia. Type 4 was proximal aplasia. Type 5 was bifocal aplasia proximally and distally. Type 6 was complete tibial agenesis with double fibulae. Type 7 was complete agenesis with a single fibula. He then assigned a score to determine the functional ability of the limb, where higher scores indicate less impairment of the limb. The tibia (0–22 points) and presence of an anlage (+10 points) weighed heaviest in importance, and the patella (0–3 points) was given slightly extra weight. The rest of the limb was scored from 0 to 2 points, and included the hip joint, distal femur, fibula, foot, and muscle function over the hip, knee, and ankle joints. Five classes were defined based on the score, indicating the degree of deficiency and difficulty of reconstruction.

The rarity of tibial hemimelia lends itself to other variants, which still may not fit any classification scheme. One reported case which does not fit any current classification is that of the intercalary deficiency, where the central portion of the tibia is missing but the proximal and distal tibia is intact [52]. The authors in this case were able to transfer the fibula into the defect with success.

Box 24.2. Jones Classification

- Type I: No visible tibia
  - Ia: Hypoplastic distal femoral epiphysis
  - Ib: Normal distal femoral epiphysis ossification (possible cartilaginous proximal tibia)
- Type II: Distal tibia deficiency
- Type III: Proximal tibia deficiency
- Type IV: Shortened tibia with distal tibia-fibula diastasis

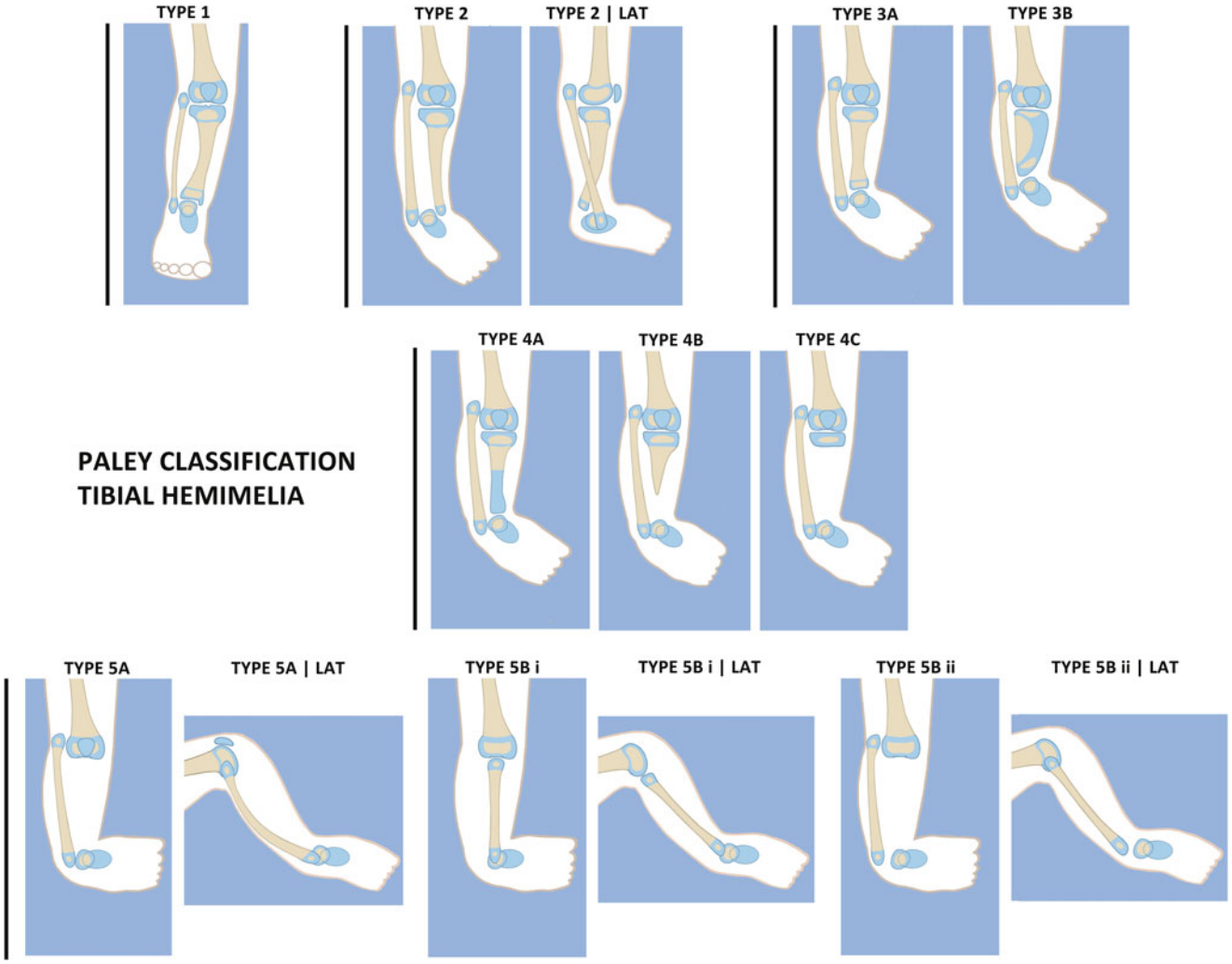

Fig. 24.2 Paley classification of tibial hemimelia

History of Treatment

Early Treatments

In 1877, Albert first published about the centralization of the fibula, fusing the femur and fibula [53]. In 1905, Myers [54] proposed a fibula-femoral arthroplasty, which was further developed by Brown in 1965. In the case of a partial absence of the tibia, there are early published reports of attempted synostosis between the tibial remnant and fibula, either side-to-side or end-to-end (Bade [55], Nove-Josserand [56]). Fraser and Robarts in 1914 reported a case with an intercalary defect in which they transplanted the contralateral fibula with reported success [57]. In 1929, Putti [58] was one of the first to give directives on treatment, describing different approaches to eight different cases: fusing the fibula to the talus in extreme equinus to increase the leg length, transfer of the proximal fibula to the intercondylar notch, and side-to-side synostosis of the fibula to the tibia both proximally and distally. Sulamaa and Ryoeppy [41] performed side-to-side opposition and recommended knee disarticulation for type I deformity.

In the current literature, the treatment of choice for tibial hemimelia has historically been biased towards amputation, though it is dependent upon the classification of the deformity. In addition, in some communities, the option of amputation is not accepted culturally, and patient families wish to pursue limb salvage. With complete absence of the tibia (Jones type Ia), most literature points towards amputation [32, 42, 48, 67, 69], while less severe deformities have several case reports of salvage options [59–61]. The presence of a tibia anlage and an active quadriceps mechanism are important factors for reconstruction, as well as prevention of knee flexion deformities [62]. Their absence may be indicated by the absence of a patella, which can be difficult

to find in young children. Thus, current imaging options such as MRI and ultrasound are a useful method of determining the presence of the patella or tibia anlage [63].

Brown Procedure (Fibular Centralization)

Brown first published his surgical procedure of fibular transfer and centralization under the femur in 1965 [64]. This procedure was done for patients with complete absence of the tibia (Jones type I). He also recommended a Syme-type amputation of the foot. In 1972, Brown published his 15-year follow-up, in which 40 of 56 patients were available for review [65]. Eighteen required secondary surgery due to a knee flexion deformity. Of the remaining 22, 1 was nonambulatory, and all but 2 were wearing braces while ambulatory. He recommended attachment of the patellar ligament to the fibula, preoperative traction, as well as femoral shortening and soft tissue releases as needed to gain extension. He also recommended surgery before age 1 for maximal ambulatory and fibular articulation potential. Inferior results were noted with an absence of quadriceps function.

Jayakumar and Eilert (1979) [66] reported six cases treated with the Brown procedure. All patients received either a Boyd or Syme amputation. Clinical outcomes were graded on several criteria of knee joint function, requiring (1) at least 10–80° of active range of motion, (2) <5° varus/valgus instability, and (3) no flexion contracture. Only two patients achieved these criteria, and they had 3+ quadriceps power. One patient had ipsilateral congenital femoral deficiency and underwent a femoral-fibula arthrodesis. Three patients had "poor" results from knee flexion contractures and two went on to knee disarticulations. The authors concluded that strong active quadriceps function of at least 3+ was necessary for Brown reconstruction to have superior results to an amputation.

Loder and Herring (1979) [67] reported nine cases of Jones type I deficiencies treated with the Brown procedure. All had preoperative knee flexion contractures. Initially, five of nine had good results with active range of motion and no flexion contractures. However, only one patient maintained active quadriceps power, and all progressively developed knee flexion contractures. Despite secondary procedures, outcomes were also poor due to knee instability and poor range of motion. At final follow-up, five had knee disarticulations and the remaining cases had poor range of knee motion (10–45°).

Epps and Schneider (1989) [68] published on three cases of Brown procedures, and noted that their results deteriorated over time due to progressive flexion contractures, and needed secondary procedures. In 1991, Epps et al. [69] reported on 14 patients (20 knees) who underwent centralization of the fibula for Jones type I tibial hemimelia. All patients developed progressive knee flexion deformities and needed multiple secondary procedures, leading the authors to recommend early disarticulation of the knee and prosthetic fitting as the treatment of choice. However, the results were considered to be failures due to the knee flexion deformities.

Simmons et al. (1996) [70] revisited the Brown procedure, publishing his results on five patients and seven limbs. All cases had Jones type Ia deformities, though one case was found to be a 1b during surgery and thus the rudimentary proximal tibia was utilized for fixation. All patients had knee flexion and varus deformities. They also had equinovarus deformities of the foot and underwent Syme amputations. An anterolateral knee incision was used, and the biceps tendon and distal insertion of the patellar tendon released. Femoral or fibular shortening was performed as needed along with peroneal nerve decompression. The fibula was centralized underneath the femoral condyles and the knee stabilized in extension with a Steinmann pin. The patellar tendon was reattached to the fibula and along leg spica cast was applied. Subsequent procedures included split thickness skin grafting, and heel pad release for the Syme amputation, and one quadricepsplasty for lack of knee flexion. All ambulated with a PTB socket with thigh extension. Average range of knee motion was 57° and only two patients achieved 10–80° of knee flexion. However, all patients were satisfied and none went on to knee disarticulation. They recommended narrowing indications for reconstruction to those patients with grade 3+/5 active quadriceps, younger than 1 year of age, no fibular bowing, and ambulatory potential.

Wada et al. (2006) [71] published nine cases of limb reconstruction in Jones type I and II cases. They performed four tibiofibular synostosis and five Brown procedures, with concurrent foot centralization procedures and subsequent lengthening as needed. The tibia-fibular fusion patients had good results with preserved knee function. Of the Brown procedures, four had poor quadriceps function and one had persistent knee instability. All five had unsatisfactory functional results based on Jayakumar's criteria [66]; however, they were all household ambulators with only one patient needing an orthosis.

Hosny (2005) [72] published a case series of 6 patients with tibial hemimelia. Amputation was not accepted in his community and they presented at an older age (3.5–13 yo). Jones type I cases were treated with gradual distraction of the fibula with an external fixator, followed by a Brown procedure 1 month later. Type II cases were treated with gradual distraction followed by fixation of the proximal fibula to the proximal tibia. Fibula lengthening was subsequently performed but had limitations due to knee flexion contractures that resulted. Femoral lengthening was then performed at a later stage. Hosny reported all patient's families were satisfied with the outcome, with all the type II patients ambulating

independently with minimal (5°) to no knee flexion contractures and active ROM greater than 90°. The type I patients could walk with a KAFO and walker and had limited ROM (10–35/40°) but could perform ADLs at home without any pain.

Overall, most authors did not report good outcomes with the Brown procedure and recommended knee disarticulation rather than reconstruction as the best option for total absence of the tibia. Many of the poor outcomes were due to progressive knee flexion contractures, knee instability, and poor range of motion, as previously defined by Jayakumar [66]. However, for some patients in whom amputation is not an option, a limb that is weight bearing though less functional may be considered a success. Certainly the presence of a strong quadriceps, patella, and proximal tibia or anlage seems to favor reconstruction, and tibiofibular synostosis has generally met with good results. In addition, the use of an external fixator prior to reconstruction can help overcome soft tissue contractures.

Amputation

Knee disarticulation has been described for treatment and remains a salvage option for failed Brown procedures, as seen in many of the studies above. Kalamachi (1985) [48] treated three children with the Brown procedure, and all went on to subsequent knee disarticulations. The failure was attributed to knee flexion contractures and no active quadriceps function, leading the authors to recommend early disarticulation of the knee without attempt of reconstruction. Alternatively, if the femur was severely hypoplastic, a femorofibular arthrodesis was performed to effectively lengthen the femur, creating a longer lever arm for improved prosthetic fitting.

Similar results and conclusions were drawn by Schoenecker et al. (1989) [32]. Of a series of 57 patients and 71 limbs, 61 limbs eventually had some type of an amputation. Brown procedures were performed on 14 Jones type Ia limbs. Ten required secondary procedures due to knee instability or flexion contractures. Based on their results, they recommended evaluating for a proximal tibia anlage to differentiate Jones types 1a and 1b, and that a type Ia should be treated by knee disarticulation due to their less-than-satisfactory results with the Brown procedure.

Fernandez et al. (1998) [38] have also been a proponent of early amputation. Twenty-two cases of tibial hemimelia were reported, of which 17 patients (and caretakers) accepted surgery. All ten Jones type I cases underwent knee disarticulation. Three Jones type II and III cases had distal amputations. Only the remaining four Jones type IV cases had conservative treatment. The author recommended early amputation so that the patient would treat it as a "congenital amputation," allowing for better adaptation to their prosthesis and rehabilitation. For many of these patients, their socioeconomic status also required a quicker and more definitive solution.

Spiegel et al. (2003) [31] described some potential complications and sequelae of amputation in patients with tibial hemimelia. They treated nine Jones type I deficiencies with knee disarticulation without any subsequent complications. Five type II deficiencies were initially treated with distal amputation (Chopart or Syme). They developed prosthetic irritation from the proximal fibula from the varus malalignment and prominent fibular head. Three patients subsequently underwent tibiofibular synostosis. One patient had progressive varus deformity and difficulty with prosthetic fitting that improved after hemiepiphysiodesis. Overall, the authors felt unsure about the best course of action for type II deficiencies. Four type III deficiencies were treated with Syme or Chopart amputations and did well.

Tibiofibular Synostosis

In the presence of a tibial anlage (Jones type Ib) or a proximal tibia (Jones type II), most authors have reported good results with tibiofibular synostosis. Kalamachi [48] reported ten patients with Jones type II tibial hemimelia that were treated with tibia and fibula synostosis. Three of these utilized a modified Brown procedure, fusing the fibula with the cartilaginous tibia anlage. They found good stability and adequate range of motion with only mild (20–30°) flexion contractures. Due to leg length discrepancies and foot deformities, half had concurrent Boyd amputations and did well with a prosthesis.

Schoenecker [32] reported 8 of 15 Jones type II limbs successfully treated with tibiofibular synostosis. Twelve were treated with a Syme amputation and were functional below-knee amputees, one had a knee disarticulation, one was nonoperative, and only one patient, who also had a tibiofibular synostosis, retained the foot. They recommended tibiofibular synostosis for Jones Ib and II with concurrent distal Syme or Boyd amputations.

Distal Tibia and Ankle Stabilization

For Jones type II deficiencies, distal tibia aplasia leads to an unstable ankle. These have historically been treated with stabilization of the ankle in the form of arthrodesis and possible amputation. In Kalamachi's [48] series, three cases had essentially normal knees, but the talus was subluxated proximally with a varus foot and prominent distal fibula. These

were treated with calcaneo-fibular fusions and Boyd amputations. Schoenecker [32] treated seven Jones type limbs with Syme or Chopart amputations to function as below-knee amputees. Tibiofibular synostosis was performed in two of these patients.

Jones type IV deficiencies have a shortened tibia with significant distal tibia and fibula diastasis. Tokmakova et al. (2003) [73] felt that the treatment of choice was reconstruction of the ankle mortise, as their patients were independent ambulators with stable ankles and plantigrade feet.

In Schoenecker's series of ten Jones type IV limbs, one limb had a Syme amputation and nine limbs had ankle joint reconstruction and salvage of the foot. Of these nine, five had a Syme amputation within 3 years due to leg length discrepancy. He recommended ankle reconstruction and leg length equalization, similar to Fernandez [38].

Limb Lengthening

Limb lengthening is also an option and adjunct for the treatment of tibial hemimelias [74]. Hootnick et al. (1977) [75] followed the natural history of tibial hemimelia and found that the leg length discrepancy remained proportional over time. Thus, they were able to calculate a final predicted leg length discrepancy to help a family decide on lengthening versus Syme amputation. They recommended amputation for predicted discrepancies greater than 8.7 cm. However, with newer technology and a better understanding of the biology, staged reconstruction and limb lengthening may allow the trained surgeon to address more severe cases of congenital tibial deficiency, preserving more lower limb function than what has been reported previously.

Desanctis et al. (1990) [76] published three cases of Jones type II deficiencies treated with staged reconstruction. They corrected the foot deformity with serial casting and posteromedial soft tissue release, followed by tibiofibular diaphyseal reconstruction, alignment of axis of the foot and leg, and limb lengthening using the Ilizarov technique. Javid (2000) [77] reported on a lengthening after centralization of the fibula in a Jones IIb deformity. They found that the fibula had delayed new bone formation, and compression-distraction technique with an Ilizarov device had to be utilized to promote osteogenesis. Devitt reported good results with differential lengthening of two cases of Weber type I deficiency [51].

Weber Patelloplasty

The Weber patelloplasty was published in 2002 [78] and described a surgical procedure in which the patella was converted into a tibial plateau. The soft tissues were gradually distracted with an external fixator to avoid the need to shorten the femur or fibula. The patella is then brought down with crossing "visor" flaps from the capsule and chondrodesed to a centralized fibula. A Z-plasty of the quadriceps tendon was used to gain length. A hinged external ring fixator was used to stabilize while allowing knee range of motion and weight bearing. A second procedure was done to achieve chondrodesis between the distal fibula and the talus. To our knowledge, there have been no publications of results of this procedure, though Weber did further describe and refine his procedure in 2006 [79].

Box 24.3. Treatment Options

- Outcomes for the Brown procedure (centralization of the fibula) depend on the presence of a quadriceps mechanism and a mobile knee without flexion contractures.
- Progressive knee flexion contractures and knee instability are common reasons for failure of the Brown procedure.
- The presence of a proximal tibia or its anlage has had good results from synostosis with the fibula.
- For some parents, a quicker and more definitive knee disarticulation may be a more attractive option, allowing a child to adapt early to a prosthesis.
- Distal tibia deficiencies can be treated with distal fusion or synostosis or Syme-type amputation.
- Advances in limb lengthening allows for limb length equalization.
- The Weber patelloplasty converts an existing patella into a proximal tibia.

New Horizons and Strategies for Reconstruction in Tibial Hemimelia

The deficiency in tibial hemimelia is a spectrum of pathology, ranging from a congenitally short tibia with relative fibular overgrowth to complete absence of the tibia. It is to be distinguished from fibular hemimelia, where there is a progressive deficiency of the fibula, ranging from a congenitally short tibia with relative fibular hypoplasia to complete absence of the fibula. An important distinction between these two conditions is that the foot and ankle are always in varus with tibial hemimelia and almost always in valgus with fibular hemimelia. The progression of deficiency of the tibia is from distal to proximal, contrary to fibular hemimelia, where

the progression of fibular deficiency is from proximal to distal. Tibial hemimelia should be distinguished from proximal deficiency of the knee, which is most commonly associated with TAR (thrombocytopenia absent radius) syndrome. Proximal deficiency of the tibia ranges from simple genu varum to a ball and socket, complete absence, or congenital fusion of the knee joint. Therefore, in order to prevent confusion, tibial hemimelia should be classified as an incremental deficiency from distal to proximal.

Most classifications in orthopedics classify pathology from the less severe to the more severe (e.g., Garden classification of hip fractures [80], Berndt and Harty classification of talar dome osteochondral fractures [81], Herring and Catterall classifications of Perthes [82, 83], and Kalamchi classification for fibular hemimelia [84]). The Paley classifications [85] for congenital femoral deficiency and fibular hemimelia (FH) follow a similar format. Some well-accepted published classifications follow the opposite format, starting with the most severe or deficient cases (e.g., Pappas classification for CFD [86], Kalamchi classification of tibial hemimelia [48], and Jones classification for tibial hemimelia [47]). The Jones classification for tibial hemimelia has been the most widely used, but it is in reverse order of deficiency. Furthermore, the Jones type III is so rare that the senior author has only seen two cases of it in 27 years of practice and more than 300 cases of tibial hemimelia. The complete absence of the proximal tibia in the Jones III makes its treatment similar to the Jones I where the proximal tibial is missing. One consideration was to add this subtype to the Paley type 5 group. I have chosen not to do this since the treatment is the same whether the distal tibia is present or not. The treatment at the knee still depends on the presence or absence of the patella and whether the fibula is autocentralized or not. The treatment of the foot does not change since the foot is in severe equino-varus as if this remnant of distal tibia was not present (Figure 24.3). Finally, the Jones classification also lacks description of several types of tibial deficiency that involve both the proximal and distal physes, such as the Delta tibia type [87] or types that have a normal appearing tibia beside a longer fibula. The Jones classification also does not consider the presence or absence of a patella.

The Weber Classification [50, 79] changed the order back to increasing severity of deficiency. It also included types with a proximal deficiency (type 3). Similar to the Pappas classification, it splits tibial hemimelia into a large number of types (total of 7). The Weber also subdivides into types that will eventually ossify and change into other types (types 3, 4, 5, 6, 7). The Weber scoring system, combined with the large number of subdivisions, makes this classification challenging for practical use.

Dissatisfied with the existing classifications and prior to Weber's publication, Paley proposed a new classification in 2003 [49]. Each type and subtype has a different surgical treatment. The progression of deficiency is from least to most, and the classification assumes that tibial hemimelia is a progressive spectrum of deficiency from distal to proximal.

Box 24.4. Paley Classification of Tibial Hemimelia (See Fig. 24.2)

- Type 1: Congenital short tibia with relative overgrowth of the fibula, distal tibial plafond intact
- Type 2: Congenital deficiency of the tibial plafond with diastasis of the tibia and fibula and relative fibular overgrowth
- Type 3: Congenital deficiency of tibia with proximal and distal physis of tibia present; distal tibial physis more dysplastic than proximal; relative fibular overgrowth
- Type 4: Absent distal tibia; proximal tibia present; relative fibular overgrowth
- Type 5: Complete absence of tibia, fibula present

Paley Classification with Subtypes and Description of Common Features

Type 1: Valgus proximal tibia (genu valgum), overgrowth of fibula proximally, plafond present and normal.

Type 2: Tibial plafond missing. Medial and lateral malleolus present; distal fibula (lateral malleolus) normally aligned to lateral talus, internally rotated with foot around the tibia. Talus is positioned between the tibia and fibula due to absence of tibial plafond. Relative fibular overgrowth present. Foot follows the fibula. The diastasis can extend as a cleft between the tibia and fibula.

Type 3a: Well-formed distal tibial physis and separate from proximal growth plate of tibia. Plafond present but dysplastic. Major relative overgrowth of fibula.

Type 3b: Delta Tibia: proximal and distal growth plates connected through bracket epiphysis. Malorientation of ankle and knee joints ends of tibia. Major relative overgrowth of fibula.

Type 4a: Delayed ossification of part, or all, of the tibia. Ankle joint present, but distal tibial physis absent. Major relative overgrowth of fibula.

Type 4b: Diaphyseal deficiency of tibia. Complete absence of distal tibia at level of diaphysis; proximal tibia has atrophic, pointed bone end often covered by separate skin pouch. Major relative overgrowth of fibula.

Type 4c: Epiphysis of proximal tibia present but absent proximal physis. Knee joint present. Delayed ossification of epiphysis. Significant relative overgrowth of fibula.

Type 5a: Complete absence of tibia: a remnant of distal tibia may be present in rare cases; patella present; flexion contracture of knee.

Type 5b: Complete absence of tibia; no patella present; flexion contracture of knee; fibula may be (i) auto-centralized or (ii) dislocated.

Limb Reconstruction Surgery

Since Brown introduced centralization of the fibula, many attempts to reconstruct the knee in the most severe types (Jones I, Paley 5) have been made. These have been met with poor results, as previously discussed. Similarly, poor results of reconstruction for Jones types II and IV and Paley types 2, 3, and 4 have led most surgeons to conclude that through-knee amputation for Jones type I, through- or below-knee amputation for Jones type II, and Syme's amputation for Jones type IV are the best treatment for each type of tibial hemimelia. In light of the advances in modern prosthetics, this is definitely a good option and should be considered as the most tried and proven method of treatment. However, advances in the treatment of all types of tibial hemimelia offer new options with excellent functional results as an alternative to amputation. The rest of this manuscript will focus on the reconstructive options for tibial hemimelia according to Paley type and subtype.

Paley Type 1 (Fig. 24.4)

These patients tend to have bilateral involvement. The condition is often familial (autosomal dominant). The tibias are

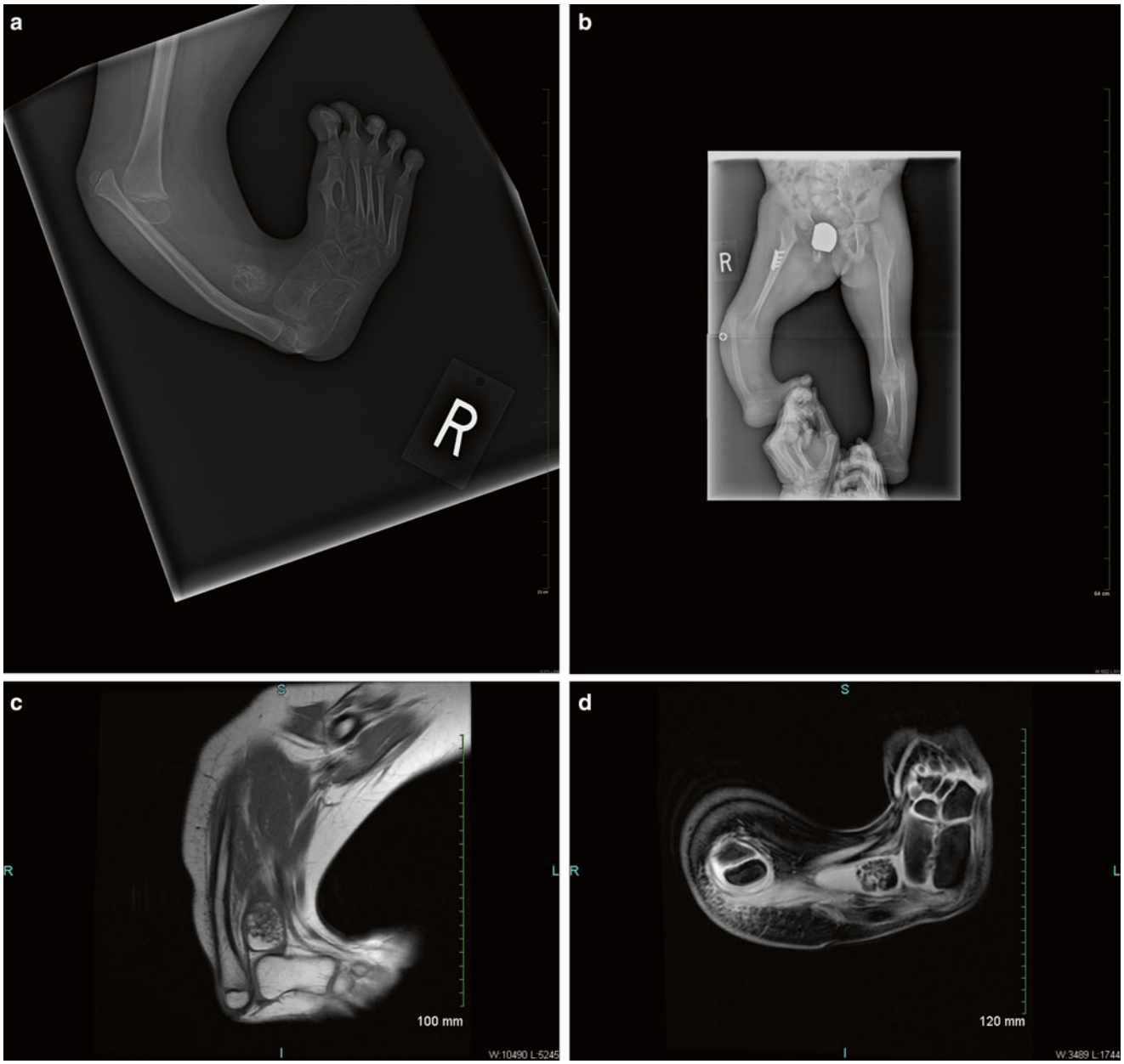

Fig. 24.3 (**a**) Jones type III tibial hemimelia. This would be classified and treated as a Paley type 5 despite the remnant of distal tibia. (**b**) Long x-ray of both legs showing the bilateral nature of this rare type. The left leg is a Paley type 3a and cannot be classified using the Jones classification. (**c**) Magnetic resonance imaging (MRI) confirming that there is no cartilaginous proximal tibia. (**d**) Another MRI cut confirming absence of the proximal tibia

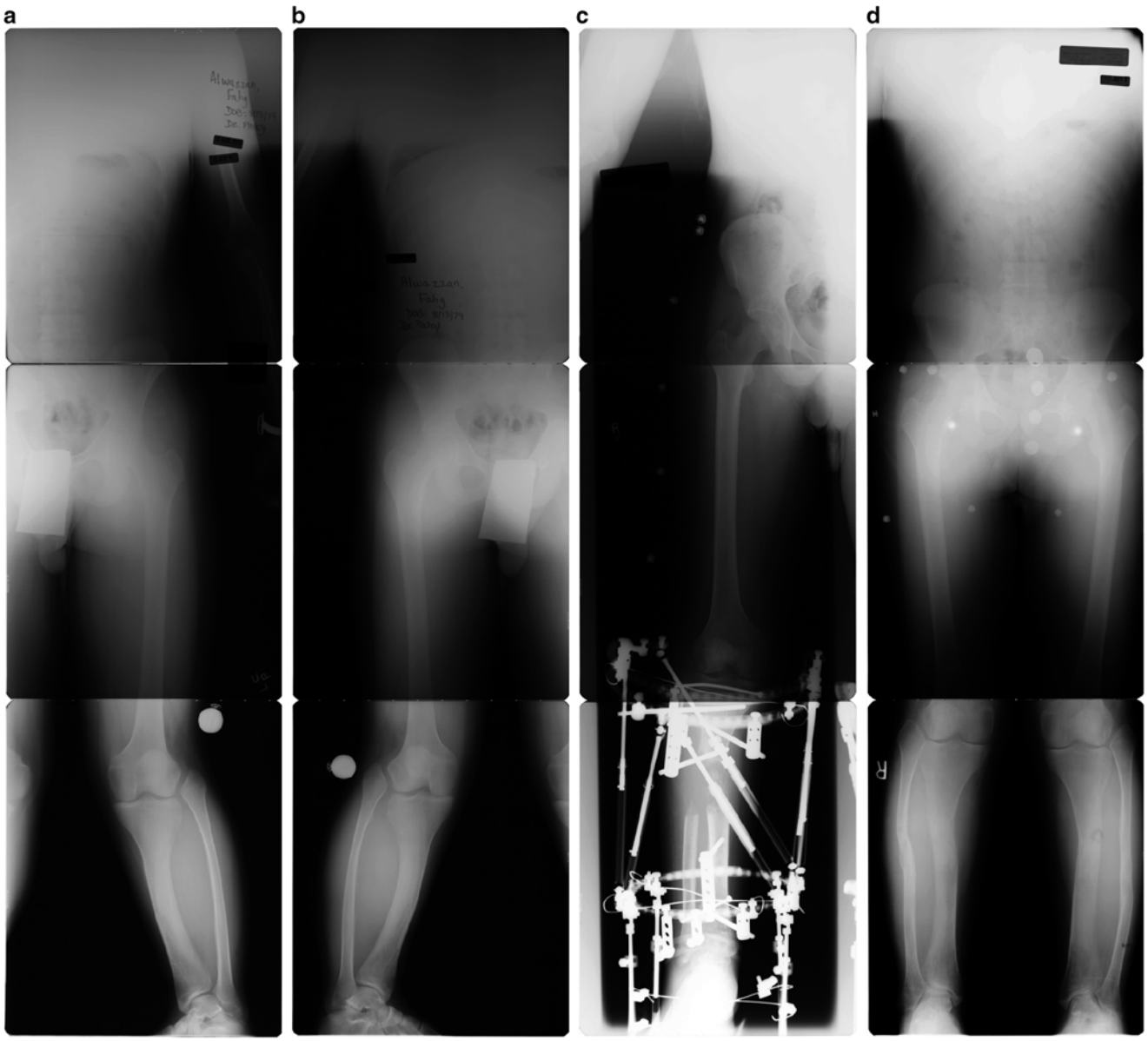

Fig. 24.4 (**a**, **b**) *Right* and *left* AP radiograph of Paley type 1 TH side showing valgus tibial deformity and overgrowth of fibula. (**c**) Lengthening and valgus correction of tibia and fibula with TSF. The fibula is not transported distally. (**d**) Final standing radiograph showing good alignment bilaterally. Proportions of femur to tibia restored closer to normal

short and the knees are in valgus. The valgus is usually from the proximal tibia, but can also be from the distal femur. If treated when the physes are open, the valgus can be corrected using temporary hemiepiphysiodesis. In adulthood, the valgus can only be treated by osteotomy. The biggest complaint of these patients is mesomelic disproportion and short stature. Therefore, the treatment is usually bilateral and includes tibial lengthening for stature in addition to correction of the valgus angulation. In regard to the overgrown proximal end of the fibula, which often articulates with the side of the femur, I prefer to leave it alone and not pull it distally. This avoids the problem of a knee flexion contracture that can result from transporting the fibula distally. If the proximal fibula is producing a noticeable bump that is bothersome to the patient, then the tibia can be lengthened without cutting the fibula, transporting the fibula distally.

Paley Type 2 (Fig. 24.5, 24.6)

The diastasis of the tibia with the fibula starting with the absence of the tibial plafond (Fig 24.4) may extend up the

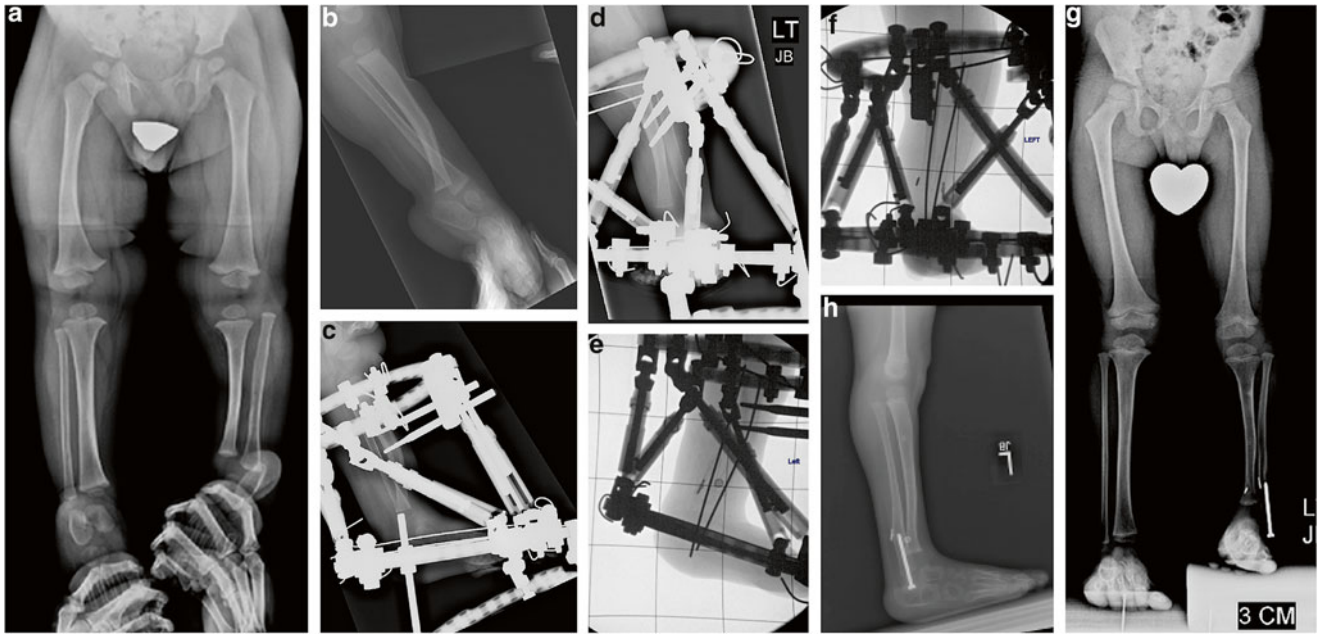

Fig. 24.5 (**a**) AP and (**b**) lateral radiographs of Paley type 2 TH. The distal fibula is more anterior than the tibia on the lateral. The distal fibula goes with the talus and is more hypertrophied than the distal tibia. (**c**, **d**) TSF device applied to foot and tibia for distraction of contracture of foot and to move the talus under the distal tibia. (**e**, **f**) After the talus is under the tibia, the ankle joint is opened and a biologic arthroplasty of the ankle joint is performed. A suture washer system is used to compress the diastasis. An intramedullary wire is placed to hold the foot in place under the tibia and the external fixator is left in place for 3 more months. (**g**, **h**) Epiphysiodesis of the distal fibula is performed to prevent distal overgrowth of the fibula. The final result is a plantigrade stable foot with some motion of the ankle

tibia. The tibia may have a varus bow to its diaphysis and in rare cases there can be a skin cleft between the two bones (Fig. 24.5). The foot always remains with the fibula. The foot is in equinovarus and internally rotated relative to the knee. The talus is proximally migrated relative to the distal tibia. The talus is at the correct level relative to the distal fibula. Since the ankle joint is to be reconstructed, the Achilles tendon is not cut but a gastrosoleus recession may be performed. The foot is repositioned by gradual distraction of the foot from the tibia using a circular ring external fixator. To prevent epiphysiolysis of the proximal and distal fibula, a 1.5-mm wire is drilled retrograde into the fibula and up the fibular diaphysis to exit through the proximal fibular epiphysis. The wire is brought through the skin proximally and then bent backwards on itself to form a hook. A small proximal incision is made and the wire is pulled back into the fibular head to lock into the proximal epiphysis. Distally, the wire is also bent 180°, then shortened and buried under the skin. This creates a temporary epiphysiodesis of the proximal and distal fibula.

One ring is applied to the proximal tibia with one wire and two half pins. The second ring is applied to the foot with three wires. The equinovarus deformity is corrected by gradual distraction of the foot away from the knee, repositioning the talus under the distal tibial epiphysis. Since the fibula is overgrown relative to the tibia, it does not need to be fixed to the distal ring. Its association with the talus and calcaneus causes it to follow the foot distally. This moves the fibula from its relatively overgrown proximal position down to the normal station.

Once the foot is located under the distal tibial epiphysis, a planned second stage surgery can be carried out. Under general anesthesia, the distal ring and wires are removed. The pin sites are covered by Betadine soaked gauzes and the leg is prepped and draped free. A transverse incision is made on the medial side at the level of the tip of the medial malleolus. The tibiotalar joint is opened, and the distal tibia and proximal talus are exposed. The tibialis posterior tendon is often found dislocated between the tibia and fibula, where the plafond should have been. It is moved out of this location and retracted postero-medial to the tibia. The distal end of the tibial cartilage is carved with a knife to the concavity of the tibial plafond, matching the convexity of the dome of the talus, creating a biologic arthroplasty. A wire, perpendicular to the sole of the foot, is passed through the dome of the talus, through the epiphysis of the distal tibia, and continues proximally into the tibial diaphysis. If the tibia has a varus diaphyseal bow to it, a percutaneous osteotomy should be made at the apex of this bow with an acute angular correction, straightening the tibia. The wire is advanced up the tibia to stabilize this osteotomy.

The tibiofibular diastasis is treated next. This is fixed by using a syndesmotic suture system such as the Arthrex-Tightrope™ or the Biomet-Ziptite™. The syndesmotic

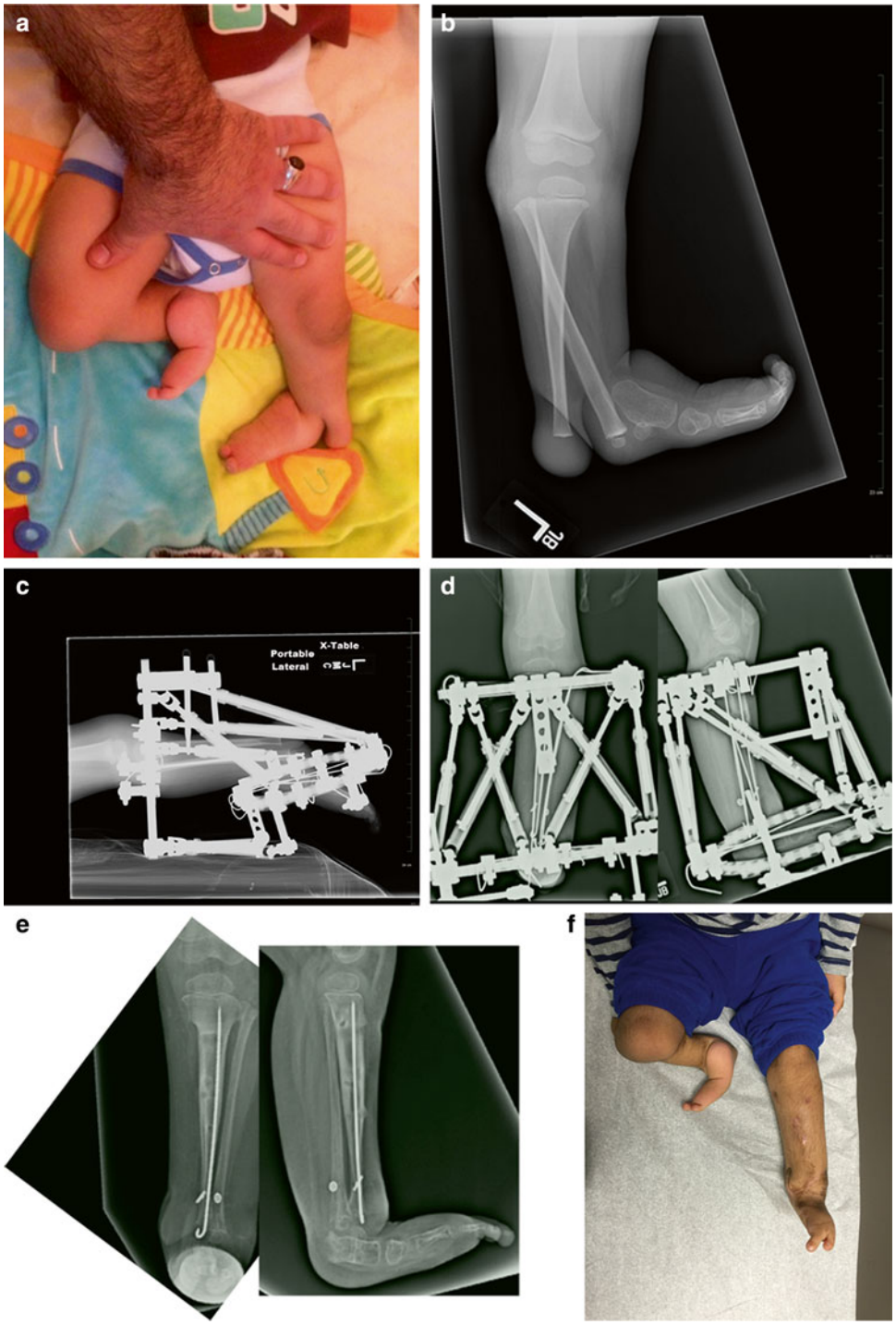

Fig. 24.6 (**a**) Paley type 2 variant: Clinical photo showing there is a cleft associated with the distal tibio-fibular diastasis. Notice that the foot goes with the fibula. There is also tibial hemimelia on the other side Paley type 5. (**b**) Radiograph showing the diastasis and cleft. (**c**) Radiograph of the TSF external fixator applied to the foot and tibia at onset of correction. Notice the 90° equinus contracture requiring the rings to be at 90° to each other. (**d**) AP and lateral radiographs after the correction is completed. The talus was centralized under the distal tibia. The syndesmotic repair was done using the Ziptite suture-washer implant. The two washers can be seen on the x-ray. The cleft was surgically closed. (**e**) Final AP and lateral radiographs showing the foot is plantigrade and the talus is centered under the tibia. The fibula is lateral to the talus. The cleft and diastasis are closed. (**f**) Final clinical photo from the front showing that the cleft is closed

suture with its two washers is used to reduce and compress across the diastasis. The incision is then closed, and the foot ring is reapplied with three new wires. This helps ensure that the foot remains in a plantigrade position.

The external fixator is left in place for 3 more months. The fibular wire as well as the transarticular tibial wire should be left in place even after fixator removal. The transarticular wire can be advanced into the calcaneus to allow for weight bearing. I prefer to leave both of these in place for 6 more months. This serves several purposes: prevention of fracture of the now osteoporotic tibia and fibula, stabilization of the ankle joint to prevent recurrence of equinus, and retardation of the faster-growing fibula to prevent recurrent relative overgrowth. Six months later, both wires should be surgically removed. A solid AFO is used until the wires are removed, after which the patient is placed into an

articulated AFO with a plantarflexion stop. Physical therapy is initiated after the transarticular ankle wire is removed.

Box 24.5. Surgical Tips

- Due to often aberrant anatomy, we recommend a mini-open incision for the Achilles tenotomy.
- A single 1.5-mm intramedullary wire protects the fibular physes at both ends and prevents epiphysiolysis.
- After the ankle arthroplasty, pass the wire antegrade through the center of the dome of the talus, then pass it retrograde through the distal tibia.
- A suture system is used to reconstruct the syndesmosis between the tibia and fibula.

Paley Type 3

The presence of a proximal and distal growth plate defines type 3. As such, the knee is usually mobile albeit often unstable due to absence of cruciate ligaments and depression or deficiency of part of the tibial plateau. The ankle plafond is present but often dysplastic, and thus does not have much motion despite its presence. The presence of the plafond differentiates it from type 2. Ankle diastasis is not typical, but some degree may be present depending on the severity of dysplastic changes of the tibial plafond.

Paley Type 3a (Fig. 24.7)

The foot is in equinovarus, and the tibia is also deformed. The circular external fixator is applied, and gradual correction of the bony deformity is carried out together with the foot, similar to the correction described for Paley type 2. At times, the foot deformity exceeds the bony deformity. After the lengthening and deformity correction of the tibia is completed, the distal tibia pins can be separated from the foot ring and connected to the proximal tibial ring. The foot and ankle are then freed to move independently with the distal ring. Once all of the deformities are corrected by distraction, the bone is allowed to consolidate and eventually the external fixator is removed. In these cases, the fibula can be treated in one of two ways: (1) resection of the mid-diaphysis of the fibula, in an attempt to create a fibular nonunion, or (2) distraction of the tibia without fixation of the fibula, to pull the fibula down to its correct station at the knee and ankle. The external fixator should always be extended to the femur to stabilize the knee joint.

Paley Type 3b (Fig. 24.8)

This type is often associated with syndromes (e.g., Werner's dysplasia). There is usually duplication of toes. This is often a familial condition (autosomal dominant) and bilateral. The bracket epiphysis can be oriented in any direction and does not always correspond to the deformity seen. The fibula is much longer than the tibia. The treatment in these cases is to consider the direction of the bracket in planning the surgery. To interrupt the bracket, the cartilage of the epiphysis and physis is cut, and an osteotomy is performed through the bone at the same level. To allow for acute correction of the deformity, part of the fibula must be resected. The acute correction is accomplished by an opening wedge osteotomy on the side of the bracket, with or without a partial closing wedge on the opposite side. The correction can be done with or without lengthening at the same time. With lengthening, it is done with the external fixator extending to the femur. Without lengthening, fixation is obtained with axial retrograde wires entering through the foot and crossing the knee.

Paley Type 4

In this type, there is a knee joint present. The degree of deficiency of the proximal tibia varies, but the knee is present and functional. The foot is in very severe equinovarus. The lack of a distal tibia makes a mobile ankle joint not possible. Articulating the talus with the distal fibula in a biologic arthroplasty has met with recurrent deformity and failure. Fusion of the talus to the distal fibular epiphysis is the goal of treatment for this type. Transfer of the fibula to the distal end of the tibia is also part of the reconstructive goal if no cartilaginous anlage exists.

Paley Type 4a (Fig. 24.9)

The salient feature in this type is the delayed ossification of part or all of the tibia. When part of the tibia is affected, it is always the distal part. An MRI is useful to image the articulations between the tibia and the femur and the tibia and the ankle joint. Based on these, a decision can be made as to whether the deformity of the tibia needs to be corrected in order to reorient the ankle to the knee. Since the fibula is longer than the tibia, two options can again be considered for managing the fibula: (1) resection to create a pseudarthrosis of the fibula, or (2) lengthening of the tibia relative to the fibula. The ankle joint, while present in these cases, is not functional. The goal is to create a plantigrade foot with a stable ankle. This usually requires distraction of the foot through the ankle joint, and then a secondary arthrotomy as described for type 2 deficiencies, to best match the articula-

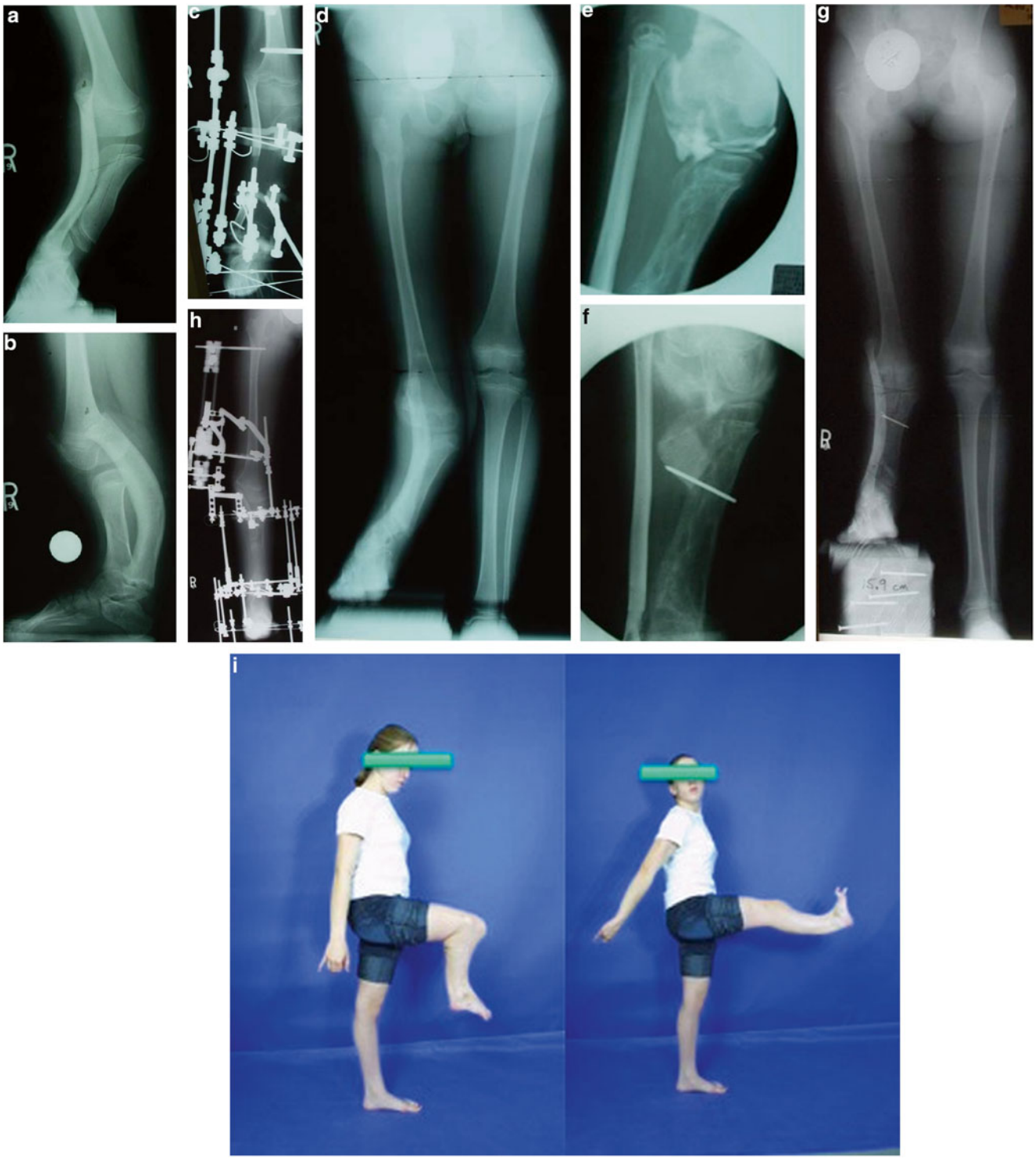

Fig. 24.7 (**a**, **b**) AP and lateral of Paley type 3a TH. (**c**) Lengthening of tibia with distal transport of fibula. (**d**) Recurrent deformity of tibia (valgus). (**e**) Arthrogram of knee showing depressed lateral plateau. (**f**) Hemi-plateau elevation to treat valgus. (**g**) Hemi-plateau elevation of lateral tibial plateau to correct the valgus and stabilize the knee. (**h**) Second lengthening. (**i**) Final result after second lengthening

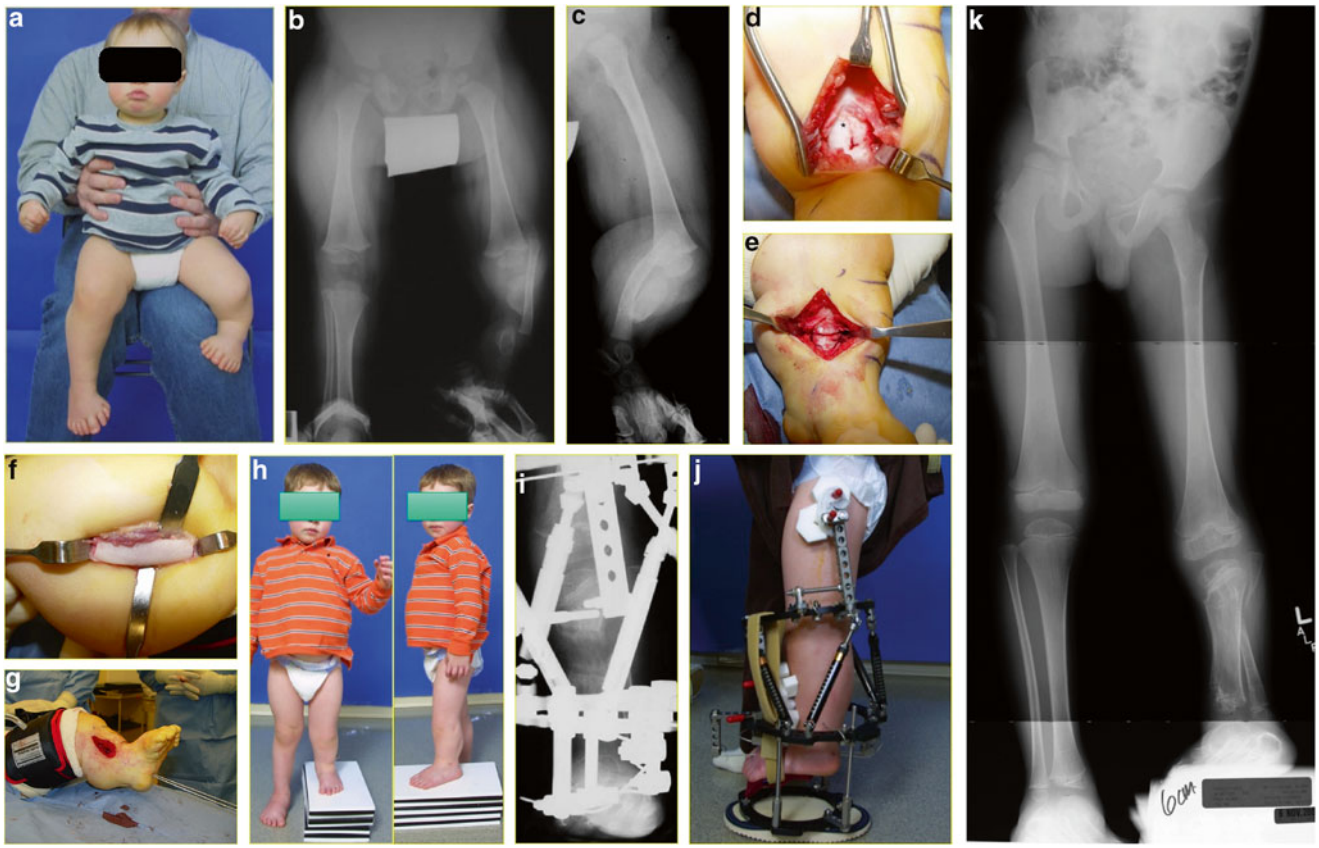

Fig. 24.8 (**a**) A 6-month-old boy with Werner's syndrome. Mother, one sibling, and grandmother have it, too. Note the syndactylized mirror hands and eight toes. (**b**, **c**) Delta left tibia Paley type 3b. The bracket epiphysis is facing anteromedial. (**d**) Cartilage of the bracket epiphysis intact (*asterisk*). (**e**) Cartilage of bracket epiphysis cut with osteotomy. (**f**) Fibulectomy. (**g**) Leg straightened by osteotomy of tibia and pinned retrograde from the foot. (**h**) After correction and amputation of extra toes. (**i**) Lengthening of tibia with external fixator to femur, tibia, and foot. (**j**) TSF device for lengthening. (**k**) Radiograph after lengthening. He will require serial staged lengthening procedures in the future to equalize leg length discrepancy by skeletal maturity

tion between the talus and tibia. The syndesmosis may or may not need to be fixed with the suture technique described for type 2. Biologic arthroplasty may also be necessary.

The unossified portion of the tibia will eventually ossify after many years. To accelerate this process, bone morphogenic protein (BMP) can be inserted into the cartilage. This is an off-label use of INFUSE (Medtronic, Memphis, Tennessee). The basis of its use in tibial hemimelia comes from the senior author's experience with congenital femoral deficiency, using BMP in a region of delayed ossification in the femoral neck [88]. Ossification of the tibia facilitates lengthening and deformity correction of the tibia through bone. If sufficient parts of the tibia are bony, an osteotomy can be made through the bony portion and pins placed in the bony portion. If an insufficient portion of the tibia is ossified to allow for external fixation, then open surgery is performed to acutely realign the foot with a tibial osteotomy, combined with resection of part of the fibular diaphysis. To ossify the tibial anlage (nonossified portion of the tibia), BMP is inserted into drill holes in the cartilage. Stabilization of the osteotomy is achieved with retrograde axial Kirschner wires through the foot and up the tibia. In most cases, ossification of the anlage is already seen by 3 months after BMP implantation surgery. Lengthening is usually done at 1 year after BMP insertion.

Paley Type 4b (Fig. 24.10)

This is the most common type 4 seen. The proximal tibial epiphysis, physis, and metaphysis are well formed, a patella is present, and active and passive knee motion are present through a normal range. The fibula is overgrown and proximally migrated and the foot is in extreme varus and, to a lesser extent, equinus. The goal of treatment is to correct the foot deformity, fuse the talus to the end of the fibular epiphysis distally, and transfer the fibular diaphysis to the distal end of the tibia proximally. This makes a one-bone leg. While this can be accomplished in one stage, it is safer, easier, and more length preserving to do this in two stages.

The first stage involves applying a circular external fixator to the femur, tibia, and foot. The knee is locked in extension by inserting an axial wire through the distal tip of the

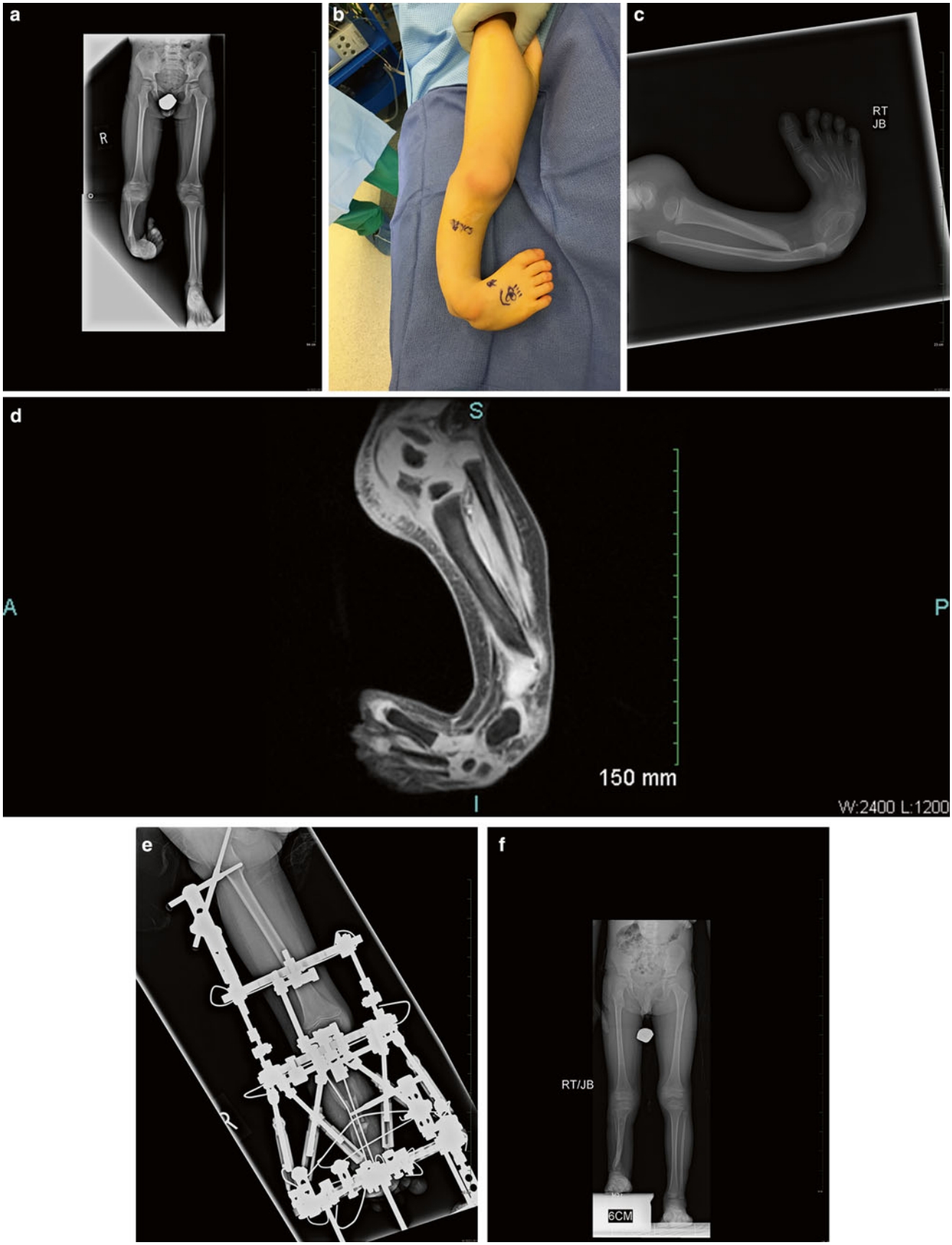

Fig. 24.9 (**a**) Preoperative AP radiograph showing Paley type 4a tibial hemimelia with severe equino-varus. Note the non-ossified region of the distal tibia. (**b**) Preoperative clinical photo showing the severe equino-varus deformity of the foot. (**c**) Preoperative lateral radiograph showing the non-ossified region of the distal tibia. (**d**) MRI showing a cartilage anlage is present articulating with the ankle joint. (**e**) A distal tibial acute angular correction combined with shortening was performed to correct the foot to a plantigrade position. The fibula was also acutely shortened. BMP was inserted into the anlage via drill holes. The tibia and fibula were gradually lengthened through a separate proximal osteotomy. The distraction gap can be seen. A femoral external fixator (to protect the knee) with hinges, is connected to a tibial external fixator for lengthening. (**f**) Final long standing AP radiograph showing the bone is straight and the distal part is ossified. In future additional lengthenings will be required for leg length equalization

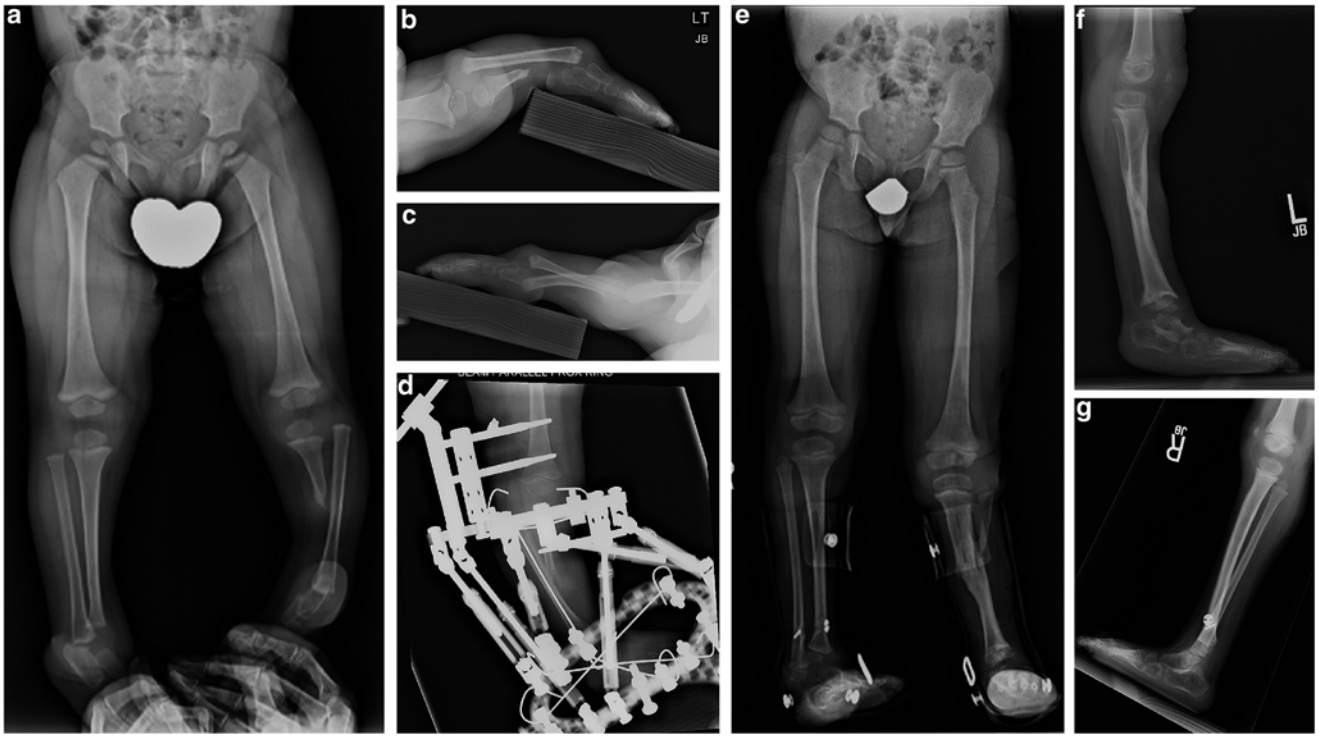

Fig. 24.10 (**a**) AP radiograph of bilateral TH with Paley type 4b on the *left* and 2 on the *right*. (**b**) Lateral radiograph of left leg Paley type 4a with severe equinovarus. (**c**) Lateral radiograph of right leg Paley type 2 with severe equinovarus. (**d**) Application of TSF to left foot and femur to correct the equinovarus deformity and bring the foot under the talus. A temporary epiphysiodesis wire is in place to prevent epiphysiolysis of the fibula during distraction. (**e**) After correction the foot was fused to the distal fibular epiphysis and the fibula was transferred to the tibia to form a one-bone leg. On the *right side* the treatment discussed in Fig. 24.4 was carried out to achieve a stable plantigrade foot. The diastasis on the right was also corrected. (**f**) Lateral of left foot. The fusion at the ankle may not be complete. The foot equinus has partially recurred. (**g**) The right foot is plantigrade and maintains some range of motion after the biologic arthroplasty

tibia, crossing the knee and inserting into the femur. The external fixation pins are in the femur only (two half pins at the femoral neck level and one wire at the ring level) and not in the tibia. This avoids having a contaminated pin site in the operative field at the time of the second stage surgery. Wires are used in the foot (three in the calcaneus and foot and one transverse in the talus). The foot is then distracted away from the tibia to correct its deformity and to gain length. This creates a large space between the talus and the end of the tibia. The goal is placing the talus distal to the lateral malleolus. To prevent epiphysiolysis of the fibula during distraction, a temporary epiphysiodesis wire is inserted into the fibula to protect its two growth plates (as described for type 2).

Once the foot is in the corrected position, the second stage surgery is carried out. The first step is to remove the foot ring and cover the pin sites with an occlusive dressing to minimize contamination during surgery. After the leg is prepped and draped free, a transverse lateral incision is made over the distal tip of the fibula. The distal epiphysis of the fibula and the dome of the talus are exposed. The capsular connections between them are cut to mobilize both bones relative to each other. A small incision is made proximally over the fibular wire. The fibular epiphysiodesis wire is cut either proximally or distally and then removed. A new straight wire is immediately inserted in the same track to protect the fibula from fracture. The fibula is often osteoporotic at this stage and without an intramedullary wire, it can easily break upon manipulation of the foot or leg. The cartilage at the end of the fibula is incised and pared back until the ossific nucleus of the fibula is reached. If there is no ossific nucleus, then BMP can be inserted to get the epiphysis to ossify. The talus is also incised parallel to the sole of the foot to the level of its ossific nucleus. The two ossific nuclei are then aligned, and the wire is advanced from the proximal end through the talus and sole of the foot to hold the foot plantigrade to the fibula.

A longitudinal anterior incision is made over the tibial diaphysis and extended distally. Fasciotomy of the anterior compartment is carried out. The anterior compartment muscles are elevated off of the tibia, and a path is then dissected to the fibula. The fibula is exposed subperiosteally. The wire in the fibula is backed out distally to the level of the planned osteotomy. The fibula is then cut at the level of the distal end of the proximal tibia. The fibula is then shifted over to the tibia. The tibia can either be split longitudinally to accept the end of the fibula inside of it, or it can be cut transversely to allow end-to-end apposition of the two bones. In both cases, the fibular wire is advanced across the proximal tibia up to the joint line of the knee. The wire in the foot is the same wire in

the fibula that is advanced across into the tibia. The external fixation wires in the foot are reinserted and connected to a ring. Struts are now used between the upper femoral ring to the distal foot ring. The external fixator maintains the alignment of the foot and knee to achieve fusion of the tibia and fibula proximally and of the fibula and talus distally. This usually takes 3 months. After that, the external fixator is removed, leaving one wire buried in the foot and fibula to protect the fibula for a few months longer. The knee motion is restored with physical therapy. In the future, lengthening of the one-bone leg can be carried out without crossing the knee joint.

Paley Type 4c (Fig. 24.11)

In this type, there is only a proximal tibial epiphysis present and no proximal tibial physis. The proximal tibial epiphysis is often unossified at an early age. The foot is in severe

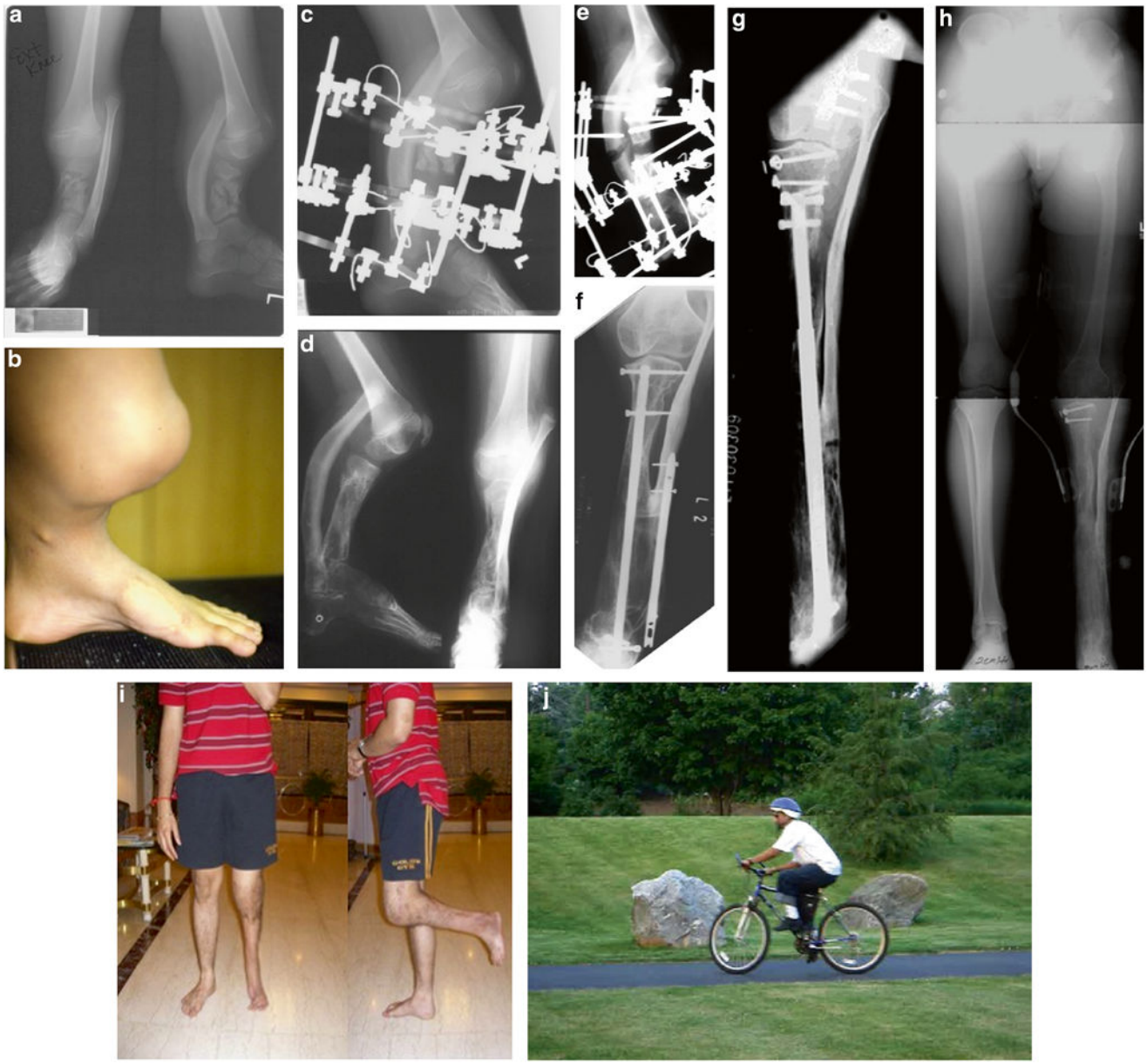

Fig. 24.11 (**a**) Radiograph of Paley type 4c. There were eight metatarsals. They were resected and transferred on a vascular pedicle to the leg to connect with the tibial epiphysis. (**b**) Clinical photo of the same. (**c**) Osteotomy across the three transplanted metatarsals and the fibula for first lengthening (6 cm). (**d**) After first lengthening and bony consolidation. (**e**) Second lengthening of tibia and fibula with Ilizarov device (8 cm). (**f**) Third lengthening over a nail in tibia and fibula (10 cm). (**g**) Fourth lengthening using ISKD internal lengthening device (8 cm). (**h**) Final radiographic result after four lengthenings and 32 cm gained. (**i**) Clinical appearance and knee function 0–90° after 32 cm of lengthening. (**j**) He is sports active, plays soccer, and rides a bicycle

Box 24.6. Synostosis and Ankle Fusion Tips

- The Paley type 4 lacks a distal tibia, and the goal is usually to obtain a fusion of the tibia remnant and the fibula proximally, and a fusion of the distal fibula and talus distally.
- The femoral ring is kept relatively high on the femur to stay out of the surgical field for subsequent surgeries.
- Protect the fibula with an intramedullary temporary epiphysiodesis wire as previously described. This is also important intraoperatively while working on either end of the fibula.
- Once the proximal fibula is brought in line with the proximal tibia, switch the transverse wires in the fibula from the distal ring to the proximal ring. Then focus on correcting the ankle deformity.
- Remove extraneous wires several days prior to surgery to minimize infection risk.
- A longitudinal split in the tibia like a clothespin allows greater surface area and healing potential.
- BMP-2 can be used to promote ossification of cartilage.
- Use the external fixator and intramedullary wires to provide stability and maintain correction until fusion is achieved.
- An arch wire can help provide compression to the talo-fibular fusion.

equinovarus, and the fibula is relatively overgrown and proximally migrated at the knee. The treatment preferred is also a two-stage surgery, with the first stage using a circular fixator to correct the foot deformity. There is often a knee flexion contracture present in these cases. It is also treated by distraction of the knee and foot with the same external fixator. A temporary fibular epiphysiodesis wire is used. A transverse wire can be added to the fibula. Initially this wire is connected to the distal foot ring, and the fibula and foot are both distracted distally, bringing the fibular head under the epiphysis of the tibia. Once this is achieved, the transverse fibular wire is detached from the foot ring and connected to the proximal tibial ring. This allows distraction of the foot deformity, and the foot is pulled distally to the end of the fibula.

In order to minimize the risk of infection, the fibular wire is removed in the office prior to going back to surgery for the second stage surgery. The same treatment is carried out to fuse the talus to the fibula as described in type 4b. At the knee, the fibula is fixed to the tibial epiphysis. If the epiphysis is unossified, then BMP is inserted into drill holes in the cartilage. The proximal fibular epiphysis is pared down as was done for the distal epiphysis. The intramedullary wire used to fix the foot is advanced proximally to transfix the proximal tibial epiphysis. Finally, the foot wire and ring are reapplied and connected with six struts to the proximal ring. It is important to use an external fixator because the axial foot wire cannot maintain the foot in a plantigrade position. The fixator remains in place for 3 months until the fibula fuses to the tibial epiphysis proximally and the talus distally. Knee range-of-motion exercises ensue after that.

An alternative form of treatment in this type is possible when there are duplicate metatarsals, which can be transplanted into the space between the tibial epiphysis and the talus. The metatarsals can be taken as free grafts, but they are preferably taken as vascular pedicle grafts flipped proximally on their pedicle. Iliac crest bone grafting between the metatarsals promotes them to unite together into one-bone mass. Once this occurs, an osteotomy across the new tibia and the fibula can be done for lengthening. The fibula remains proximally dislocated as long as it does not impede knee motion. Repeated lengthenings are needed to equalize limb length discrepancy.

Paley Type 5

Complete absence of the tibia presents the biggest challenge for reconstruction because there is no knee joint. While ankle fusion gives good function with little disability, knee fusion leads to significant disability for sitting and climbing stairs. It is preferable to avoid a knee fusion. Even if active knee motion cannot be achieved, a mobile knee joint supported by a brace is preferable to a knee fusion. This is not dissimilar to a paralytic knee from polio. Therefore, the following two methods have been developed to reconstruct the knee.

Paley Type 5a (Fig. 24.12)

If a patella is present, it can be converted to a tibial plateau. This original idea was first published by Michael Weber [78, 79]. The patella is moved on a vascular pedicle from its normal position, anterior to the femur, to the distal end of the femur. The fibula is centralized to the patella and its epiphysis fused to the patella. This procedure is referred to as the Weber patellar arthroplasty or Weber procedure.

The Weber procedure can be performed acutely (as described by Weber) with gradual correction of a flexion contracture of the knee joint, using a hinged external fixator, or following staged gradual distraction of the fibula to first correct the knee flexion contracture (according to Paley). When performed acutely, the foot deformity is gradually distracted at the same time as the gradual knee contracture correction. When performed after the distraction, the foot is distracted away from the fibula as described above with a temporary epiphysiodesis wire in place. Once the femur,

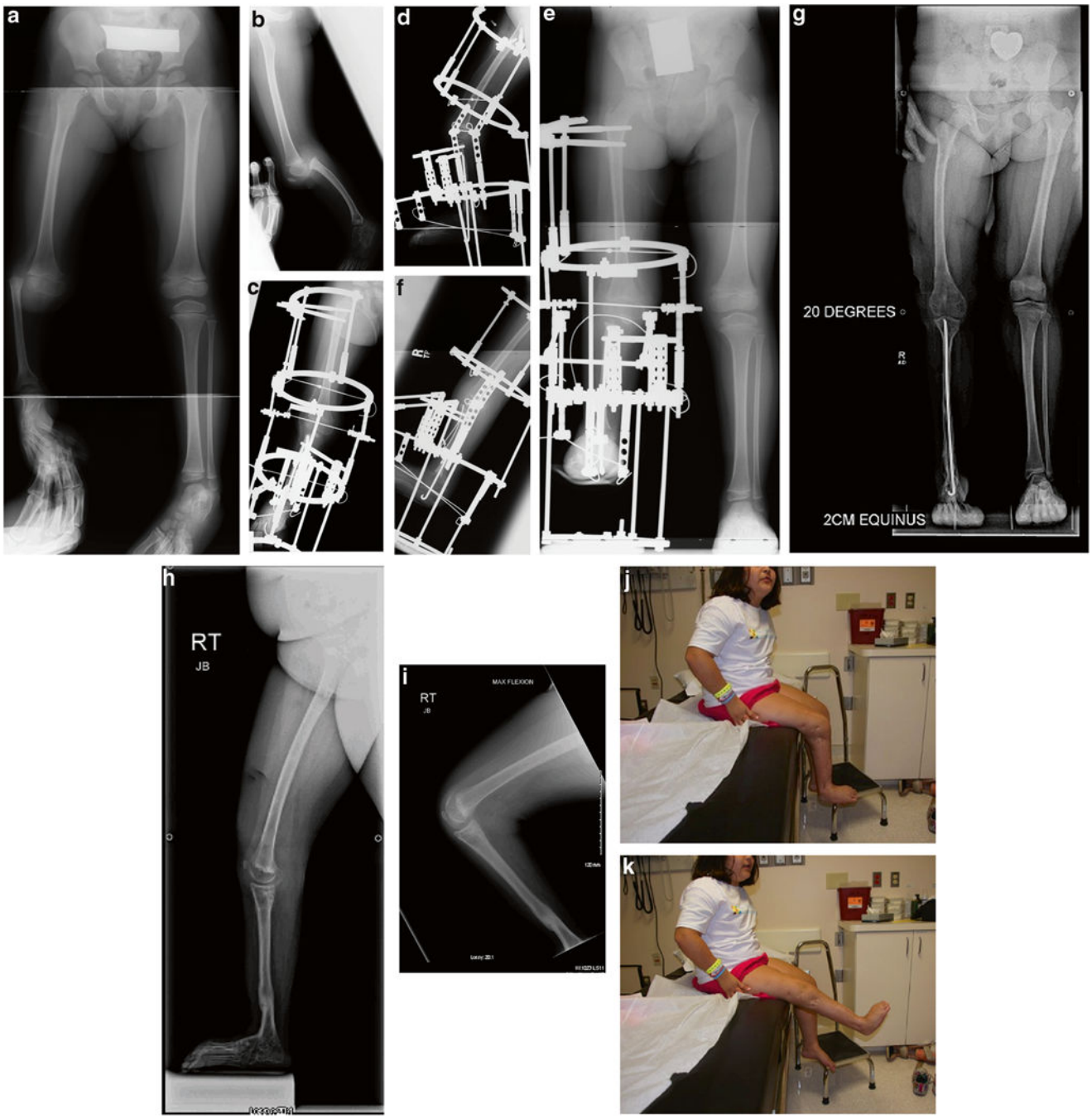

Fig. 24.12 (**a**, **b**) AP and lateral radiographs of Paley type 5a TH. The foot was previously treated and remains in an equinovarus deformity. (**c**, **d**) AP and lateral radiographs after the Weber patellar arthroplasty was performed and the patella jointed to the fibula. The ankle was fused in a plantigrade position for the foot. Initially there was a flexion contracture of the knee. The circular external fixator was used to correct this. (**e**, **f**) After distraction the knee is straight. (**g**) After removal of the fixator an intramedullary wire was used to help protect the fibula. (**h**, **i**) AP and lateral of the knee and tibia after healing from the Weber. Note how well remodeled the knee appears. Due to leaving a small fragment of the patella behind a new patella developed. The knee has excellent active range of motion. (**j**, **k**) Clinical photos of the leg after one lengthening. Note the active knee flexion and extension

fibula, and foot are all aligned, the Weber procedure can be performed through a longitudinal anterior incision over the distal femur and proximal fibula. At the distal end, the ankle fusion can be performed as described above through a transverse lateral incision.

Weber Patellar Arthroplasty (Fig. 24.13)

An anterior midline incision is made over the distal femur and proximal fibula. Medial and lateral skin flaps are elevated to expose the fibula and distal femur. The patella is

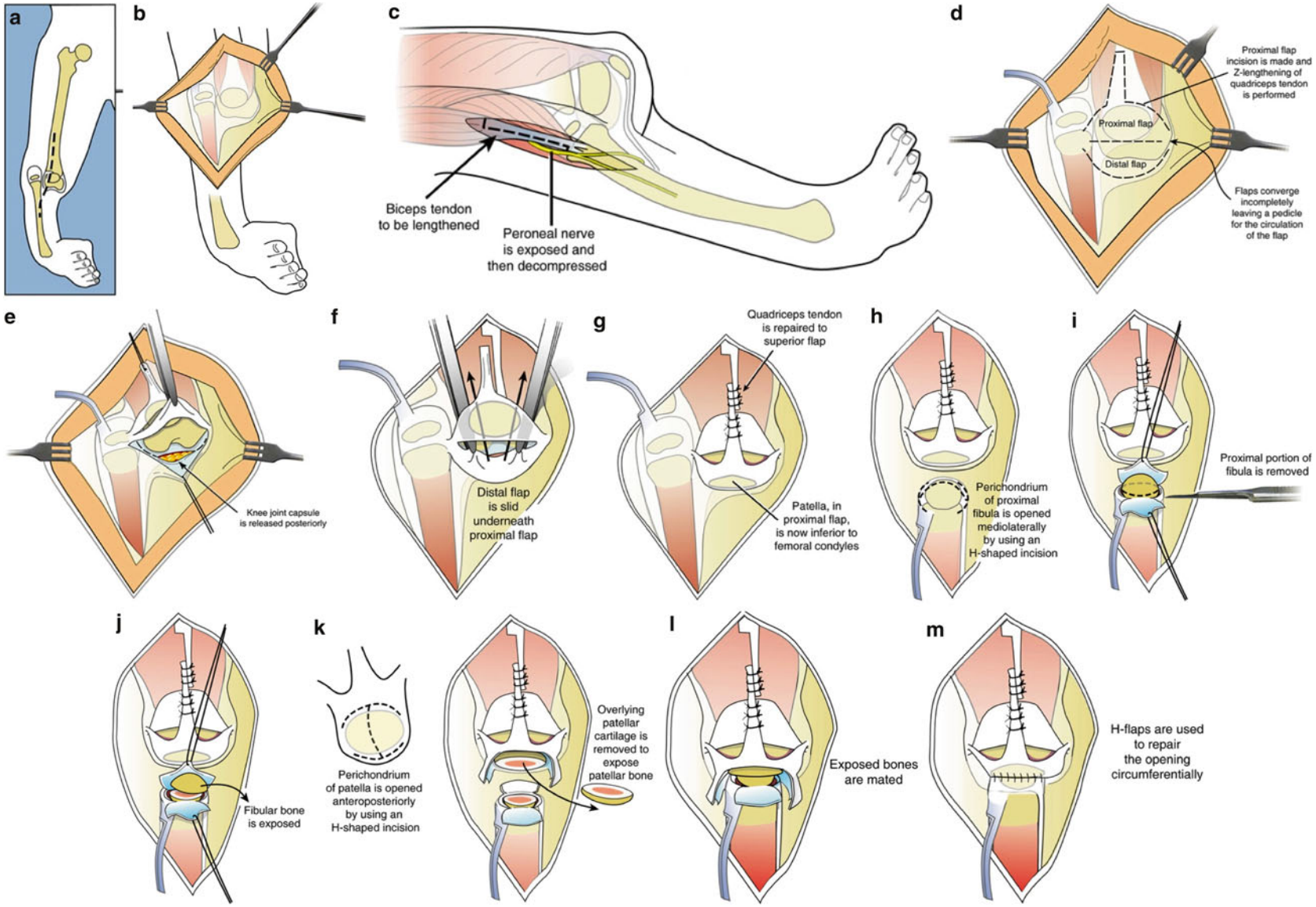

Fig. 24.13 (**a**) Weber patellar arthroplasty incision. (**b**) The underlying fascia is exposed over the patella, femur, and tibia. (**c**) The peroneal nerve should be exposed and decompressed and the biceps tendon lengthened. (**d**) The two visor flaps are marked out. (**e**) The visor flaps are incised and the posterior capsule is cut with great care to identify the vessels posteriorly so as not to injure them. (**f**) The two visor flaps are flipped so that the proximal one goes distal and the distal one goes proximal. (**g**) The quadriceps tendon is repaired and the visor flaps sutured together. (**h–k**) Corresponding H-flaps are incised into the perichondrium of the patella and the fibular head and the ossific nucleus of the fibular head and the patella exposed by resecting some of the overlying cartilage. (**l**) The patella and fibular head are mated to each other. (**m**) The H-flaps are sutured together

Box 24.7. Weber Patelloplasty Tips

- Correct knee flexion contractures as much as possible with frame distraction before surgery. A posterior capsulotomy may be necessary.
- Find and protect the peroneal nerves and the posterior tibial neurovascular bundle.
- If there are remnants of the medial or lateral hamstrings, these can be preserved and reattached to the fibula at the end of the operation.
- Keep the visor flap pedicles as wide as possible. Deepen the cuts just enough to allow the mobility to slide them around each other. Usually the upper patella flap goes underneath.
- Patellas can be present in various shapes. A vertical patella can be easily split, such that a small remnant is translated superiorly with the visor flap and form a new patella.
- The H-flaps should be vertical on one side and horizontal on the other and should be determined by the anatomy and shape of the patella and fibula.
- Cut down into the cartilage of the patella and proximal fibula until reaching the ossific nucleus. If these are not present, then consider placing BMP-2 to promote ossification.
- Suture the posterior H-flap first before fixing the patella and fibula with a wire.

identified. Two "visor" flaps are outlined with a marking pen. At the medial and lateral ends, the pedicle for each "visor" is kept as wide as possible. The proximal visor flap contains the patella. The distal visor flap is all capsular. In the Paley modification, the distal visor flap includes a small part of cartilage of the inferior pole of the patella. This allows a new patella to form anterior to the femur. A Z-lengthening of the quadriceps tendon is performed as part of the proximal visor flap. If the fibula or patella is unossified, Weber recommends using two perichondral H-flaps to get the two cartilage surfaces to heal together. To ensure bone-to-bone healing, we prefer to insert BMP into drill holes in the cartilage of the patella and fibular epiphysis. To avoid injury to the popliteal vessels, it is preferable to find and protect them before completing the posterior capsulotomy.

Since the knee and ankle were predistracted, they can both be fixed by one axial wire across the knee joint. The circular external fixator is used only as a holding device to ensure full knee extension and a plantigrade foot. The medial and lateral collateral ligaments should be reconstructed using local tissues (biceps tendon laterally and semitendinosus medially). After 3 months, the external fixator is removed and the trans-knee wire shortened to span only the fibula and foot. After the external fixator is removed, the patient goes into a cast for a month; when the cast is removed, they are fitted for a KAFO brace. A brace is needed for many years until the knee is stable enough to allow walking without the brace. This is usually after age 10, when adequate hypertrophy of the joint surface and fibula has occurred.

Type 5bi (Fig. 24.14)

If there is no patella, but the fibula is autocentralized, then there is usually a quadriceps muscle in continuity to the fibula. The distal femur is usually less dysplastic in these cases. The knee still presents with a fixed flexion contracture and the foot presents as with type 5a, dislocated and in extreme varus and equinus. The treatment is to distract the knee contracture until the fibula and femur are collinear with each other. The foot should also be distracted relative to the fibula to centralize it under the distal end of the fibula. This is accomplished with an Ilizarov device on the femur using hinges and a circular ring fixator for the foot correction. This permits simultaneous correction at both ends of the bone. This is possible because the fibula is autocentralized. If the fibula is dislocated as in type 5bii, a circular fixator is needed for both knee and ankle and the correction is done sequentially as for type 5a. Once the distraction correction at both knee and ankle is completed, a second stage surgery is performed to reconstruct collateral ligaments at the knee and to advance the quadriceps muscles onto the fibula. An ankle fusion as described above is performed. The distal ring on the foot is removed and then reapplied in this surgery. All wires near the knee are removed in the office at least 1 week prior to the surgery. The external fixator is left in place for another 3 months and then removed. A spica cast is applied for a month and then an HKAFO brace is made with a hinged knee and solid ankle.

Type 5bii (Fig. 24.15)

If there is no patella and the fibula is dislocated, the fibula can be centralized by distraction. First, a temporary epiphysiodesis wire is inserted up the fibula and hooked over each epiphysis at both ends. This is to protect both proximal and distal fibular physes. An Achilles tendon tenotomy is carried out. Then, one proximal and one distal circular fixator ring is applied with two half pins in the proximal femur (one up the femoral neck and one transverse at the level of the lesser trochanter) and one wire on the proximal ring in the mid-femur. A distal ring on the foot is applied with three anterior posterior wires in the foot (one from posterior midcalcaneus to anterior mid-forefoot, one from posteromedial calcaneus to anterolateral forefoot, and one

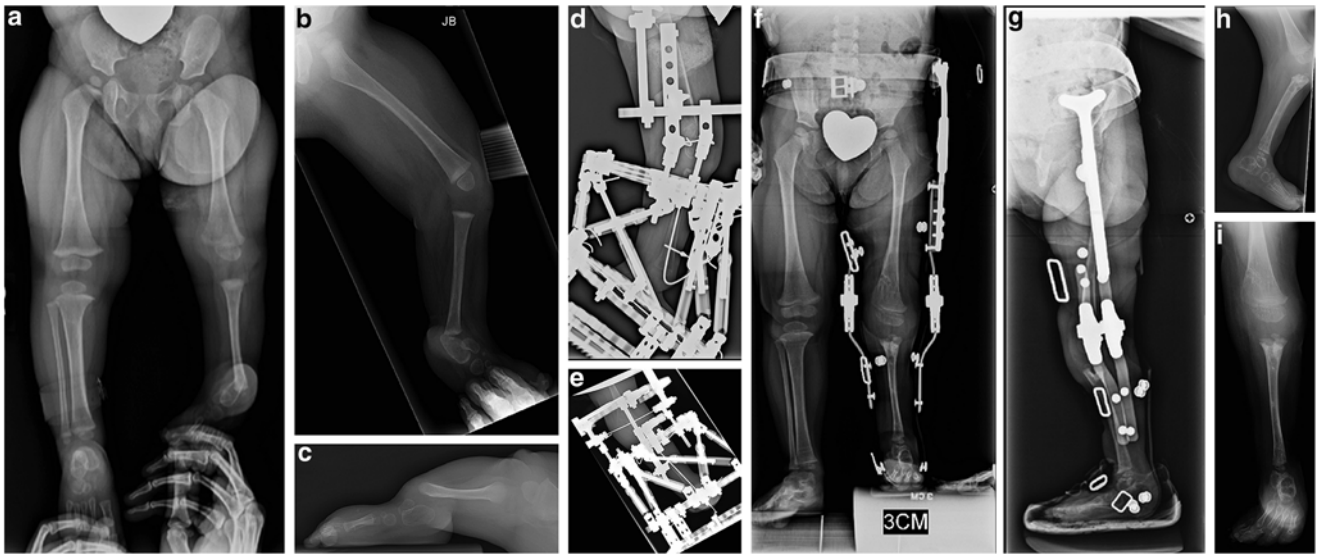

Fig. 24.14 (**a**, **b**) AP and lateral radiographs of Paley type 5b (i) TH. The fibular is autocentralized on the femur. For this reason it has hypertrophied. The knee joint has a fixed flexion deformity. (**c**) The foot is in severe equinovarus relative to the fibula. (**d**, **e**) Hinged construct used to correct knee contracture. A knee hinge wire is located across the distal femur. Taylor Spatial Frame is used to correct foot contracture gradually. A temporary epiphysiodesis wire is in place in the fibula to prevent epiphysiolysis. (**f**, **g**) After the knee and foot are corrected, the knee is reconstructed by open surgery to do a quadricepsplasty and to reconstruct the collateral ligaments. The ankle is fused: talo-fibular fusion. The external fixator is left on for 3 months after this second surgery. It is then removed and a brace is used to support the leg. (**h**, **i**) The distal fibular epiphysis is fused to the talus in a plantigrade position. The knee is well reconstructed

from posterolateral calcaneus to anteromedial forefoot) and one transverse wire across the ossific nucleus of the talus. One wire is inserted into the fibula and connected to the foot ring. The spatial frame planning is done relative to the proximal ring for the knee joint contractures. Once the fibula is reduced at the knee and the knee flexion contracture eliminated, the fibular wire is connected to the proximal ring and the spatial frame planning repeated for the distal ring and foot contractures. When both ends of the fibula are aligned to the knee and ankle, the second stage surgery can be performed.

Paley Centralization of the Fibula on the Femur (Fig. 24.16)

Through a midline anterior incision, the distal femur and the proximal fibula are exposed. The peroneal nerve is liberated and decompressed from the fibula. The biceps tendon is split with half of its tendon left attached to the fibula to be used as a lateral collateral ligament. The other part is transferred to the fibula anteriorly to rebuild a quadriceps muscle. On the medial side, the semitendinosus muscle is also transferred anteriorly. The tensor fascia lata and its iliotibial band are also harvested and transferred. The adductor magnus is also transferred. These four muscles are connected to the quadriceps muscle. The quadriceps muscle usually ends in the mid-thigh and does not continue to the knee. The transferred muscles are all connected together with the lateral and medial muscles balancing each other and centralizing themselves to the iliotibial band, which acts as a central rib for the connection of all of these muscles. It is eventually attached to the front of the fibula to act as the patellar tendon.

Before connecting the transferred muscle, it is important to anchor the fibular head to the femur. This is done by creating an interosseous ligament between the femur and the fibula. One can use allograft or the half of the biceps tendon for this purpose. This single step prevents subluxation of the fibula from the femur. As dislocation was the main reason for failure of the original Brown procedure, the interosseous ligament prevents subluxation from occurring while permitting hinge flexion of the two bones. The ankle is fused as previously described. A wire is inserted from the foot through the fibula and then through the knee joint. The foot ring, which was removed at the start of the procedure, is reapplied with the circular fixator struts. The foot is immobilized in a plantigrade position and the knee in full extension. Three months later, the external fixator is removed, but the medullary wire crossing the ankle and knee is left in place. A cast is used for 1 month and then a KAFO is prepared. The wire is removed 6 months later and knee rehabilitation is started. Other than amputation, the alternative or fall back operation for this type of tibial hemimelia would be a knee fusion. In unilateral cases, this is a reasonable alternative.

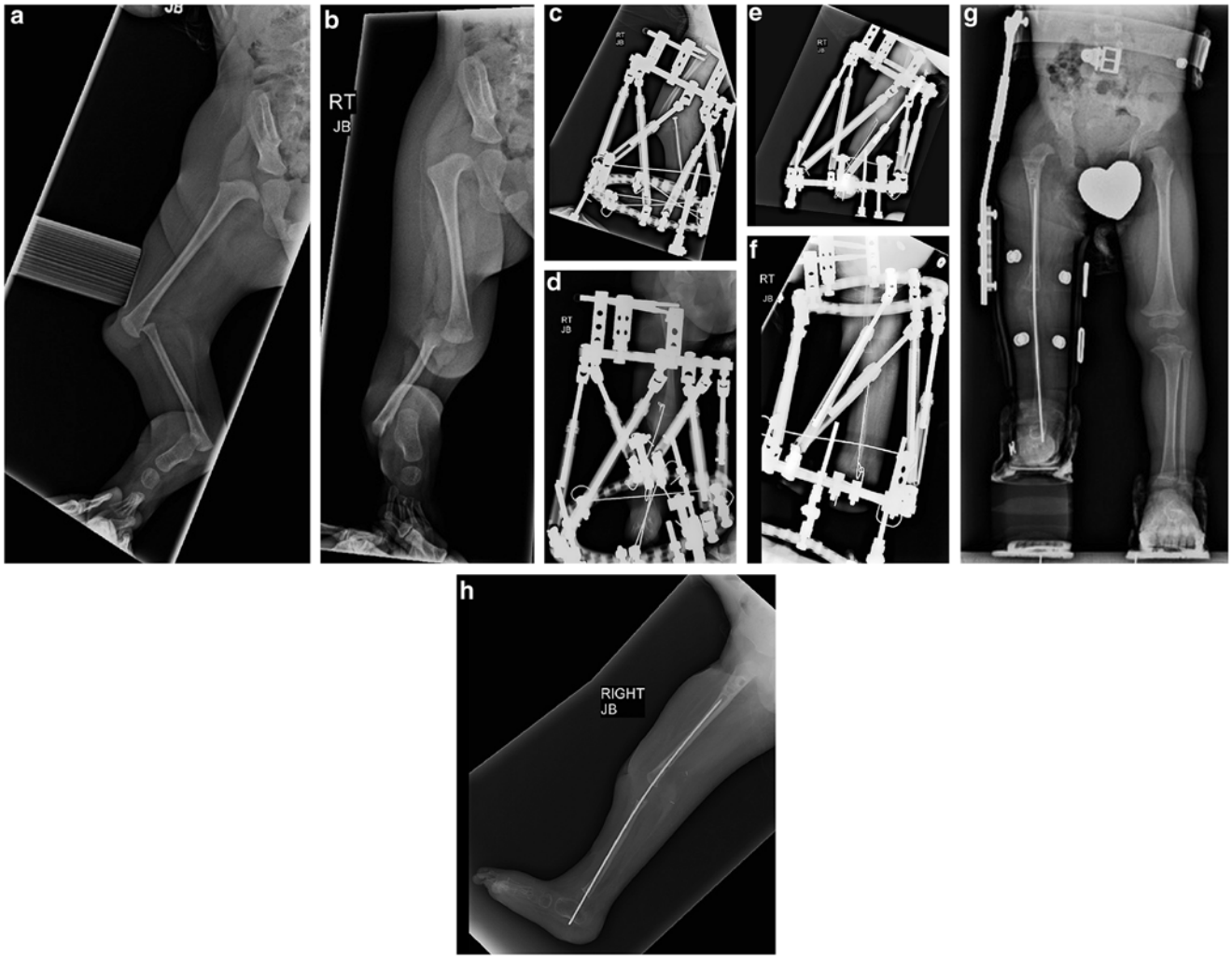

Fig. 24.15 (**a**, **b**) Radiograph of Paley type 5b (ii). There is a dislocation of the fibula on the femur and the foot on the fibula. There is a severe equinovarus of the foot and flexion contracture of the knee. (**c**, **d**) A Taylor Spatial Frame is applied from the femur to the foot with one wire across the tibia. A temporary epiphysiodesis wire is in place in the fibula. (**e**, **f**) After 3 months of distraction the knee contracture is eliminated and the foot is reduced below the end of the distal fibula. (**g**, **h**) The foot is fused to the distal epiphysis of the fibula and the biceps, semitendinosus, tensor fascia lata, and adductor magnus muscles are all transferred to the knee to substitute for the missing quadriceps muscles. A temporary arthrodesis wire is placed across the knee and ankle. The external fixator was removed 3 months after the knee-ankle surgery. The wire across the knee and ankle is left in place for 6 months to allow better loading and minimize the risk of recurrence. It is then removed

Summary

Through-knee amputation remains the most commonly used procedure for complete tibial aplasia. It is certainly a reliable option that can be carried out by most orthopedic surgeons. It should be seriously considered in unilateral cases. In bilateral cases of Paley type 5, complete tibial aplasia reconstruction by one of the techniques described above should be considered, especially on the side that has a patella.

Amputation is probably overused in Paley type 2, 3, and 4 tibial hemimelia. In the senior author's opinion, reconstructive results for these types are reliable and successful in achieving a functional lower extremity. For type 5a, where a patella is present, the results of a Weber patellar arthroplasty in the hands of experienced surgeons are also very satisfactory. The reconstructive options for tibial hemimelia have improved greatly over the past two decades. With continued success and improvements, they may one day overtake the amputation option. Other anatomic variants of tibial hemimelia including duplication of fibula, tarsals, metatarsals and toes, and secondary femoral condyles, have not been discussed. They are not included in the classification either. These associated anatomic variants must be considered in the treatment but are beyond the scope of this chapter.

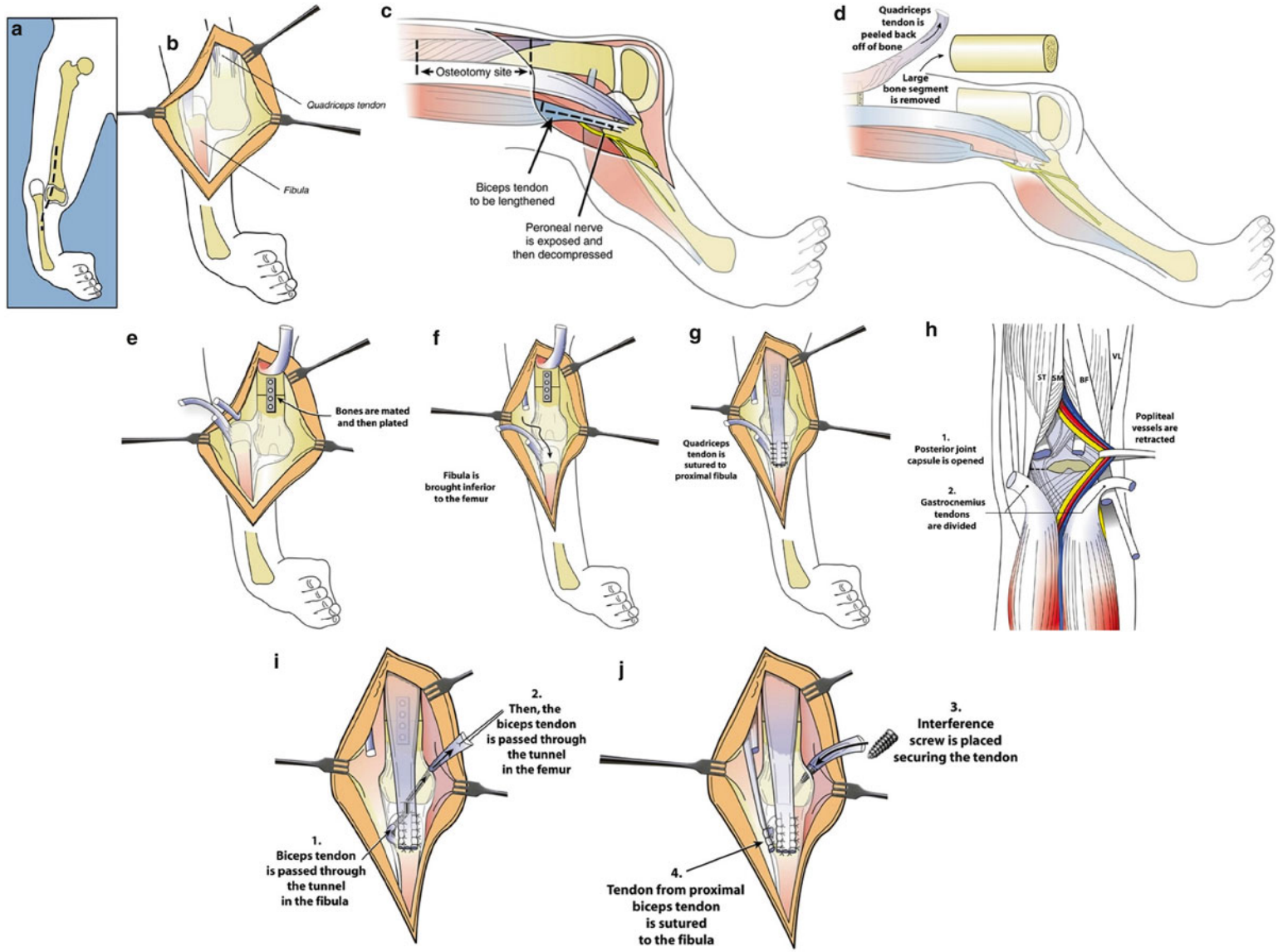

Fig. 24.16 (**a**, **b**) Incision for acute reduction of Paley type 5b (ii). (**c**, **d**) Biceps tendon lengthening, peroneal nerve decompression, and femoral shortening to treat the knee flexion contracture. (**e**) Fixation of the distal femur. (**f**) Reduction of fibula to femur. (**g**) Advancement of the remnant of quadriceps to the fibula. (**h**) To fully correct the knee flexion contracture, the vessels are identified and the posterior capsule is incised. All of the hamstrings and gastrocnemius tendons are released. (**i**) The biceps tendon is used to create a cruciate ligament to tether the fibula to the femur. (**j**) The biceps is advanced and the new cruciate ligament is secured

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