Case 3: 2-Year-Old with Neurofibromatosis and New Onset Fracture

Presenter: Kenneth Noonan MD, MHCDS (KN)
Expert Panel: Christopher Iobst MD (CI); Hitesh Shah, MBBS, MS (HS)
Moderator: Jennifer C. Laine MD, (JL)

Brief History: This was a 15-month-old girl who had a right tibial deformity since birth. Her mother has a history of neurofibromatosis and our patient has multiple café-au-lait spots. She was in her usual state of health when she fell from a stool and had acute pain in her leg. The x-ray to the right was taken in the emergency department (Figure 1).

She was treated in a long leg cast for 3 months and her fracture had not healed. (Figure 2).

Decision Point #1—Expert Panel

JL: Doctor Iobst and Professor Shah, at this point, what is the problem list for this patient?

CI and HS:
- Tibial pseudarthrosis
- Angular deformity of the tibia in both planes
- Dysplastic bone
- Abnormal periosteum
- Leg length discrepancy
- Secondary deformity of the ankle

JL: What are your concerns for this patient?

HS: With conservative treatment, the fracture will not unite. Sclerosis around the fracture in the tibia is the main concern for surgical intervention. Deformity in the sagittal plane is another concern for fixation with intramedullary (IM) nailing. Shortening would be an issue with excision of the sclerotic part of the tibia. The intact fibula must be treated with osteotomy to avoid persistent distraction at the fracture site of the tibia.

CI: Conservative management will not work. Surgical intervention will be required.

JL: If this were your patient, what would you recommend and why?

CI: Surgical intervention, including:
- Resection of the pseudarthrosis
- Acute shortening of the tibia/fibula

Figure 1. A nondisplaced fracture is now seen in her anterolaterally bowed tibia.
Figure 2. Three months after fracture, her tibia has not gone on to heal and the fracture site appears more dysplastic.
• Bone grafting to promote cross-union (to increase surface area of healing)
• Circular external fixator using only wires and proximal osteotomy for restoring length

**HS:** I would recommend excision of the pseudarthrosis of the tibia (until fresh bleeding), osteotomy of the intact fibula, IM nailing of tibia and fibula, synostosis between distal tibia and fibula, excision of diseased periosteous, and cortical bone grafting from the contralateral tibia. These recommendations are for the union of the distal tibia and fibula and to prevent the progression of the deformity. The cortical bone graft resists the osteoclastic resorption.

**Treatment for Case 3**
At 20 months, she was treated with resection of the pseudarthrosis, iliac crest bone grafting, and use of BMP-2 sponges at the pseudarthrosis site. She was stabilized with IM fixation of the fibula with a K-wire and retrograde nailing of the tibia with a 5.5 stainless steel scoliosis rod (Figure 3). At 3 months, she showed signs of healing, but the rod had migrated distally (Figure 3b and 3c). Based on rod migration we realized she could not grow off of the rod which would transfix the ankle; thus, she underwent surgery, where we modified the spine rod to accommodate a 2.7 mm stainless steel screw. Her pseudarthrosis site was re-bone grafted with more autograft and another BMP-2 sponge (Figure 4).

At 5 years of age, she had had developed discomfort associated with the cross-locking screw in the tibial nail as well as at the ankle from the migrating fibular K-wire. After discussion of the risks, benefits, and alternatives, the parents elected to proceed with removal of the tibial screw and fibular rod (Figure 5).

At 6 years of age, she was developing distal tibia valgus, and she underwent guided growth of the medial distal tibia (Figure 6).

**Figure 3a (left).** The hamartoma prior to resection and stabilization with a k-wire in the fibula and a 5.5 rod in the tibia and foot. **Figure 3b (middle) and 3c (right).** The pseudarthrosis appears to be healing but the rod has migrated distally.

**Figure 4.** One month after the rod revision. She is now 2 years old with bridging bone and is further protected with this AFO.

**Figure 5.** At 5 years of age, our patient had pain over her cross-link screw as well as her distal fibula k-wire. Both were removed.

**Figure 6.** Placement of a medial screw was performed in order to decrease her ankle valgus.
At 7 and half years of age, she had developed some deformity of her tibia distal to her rod and the metal tip was now painful (Figure 7).

**Figure 7.** The distal end of the rod had migrated through her tibia, and she was painful.

### Decision Point #2—Expert Panel

**JL:** Doctor Iobst and Professor Shah, what are the key problems at this point?

**CI:**
- Multiplanar tibial deformity
- Fibular pseudarthrosis
- Painful prominent hardware
- Leg length discrepancy

**HS:**
- Procurvatum deformity of the upper third-middle third junction and middle third-lower third junction
- Valgus deformity of the tibia
- Valgus deformity of the ankle
- Fibula thin with hypoplasia and pseudarthrosis
- Unprotected segment of tibia above the rod and below the rod (upper third and lower third of the tibia).

**JL:** What are your concerns for this patient?

**HS and CI:** The tip of the rod is outside the bone. With the lysis around the rod distally, there is an impending fracture of the lower third of the tibia. A revision of the rod might be possible only after osteotomy of the tibia (that might cause delay in the union at the osteotomy site). Bone grafting would be needed at the osteotomy site. Pain at the tip must be evaluated to rule out fracture and infection. The ankle is also probably stiff from the implant being placed through the tibio-talar joint.

**JL:** If this were your patient, what would you recommend, and why?

**CI and HS:** Complete blood count, ESR, CRP. Oblique radiograph to rule out re-fracture (if in doubt, CT scan to rule out re-fracture). Removal of the prominent painful hardware, tibial osteotomy, insertion of a SLIM™ nail (or half of a Fassier-Duval [FD] rod) in the tibia and fibula, bone grafting to promote cross-union between tibia and fibula. These recommendations are for the union of the distal tibia and fibula and to correct the progression of the deformity.

### Continued Treatment of Case 3

Our team was worried about impending fracture, and we advised the family to consider rod removal and the use of flexible nails to maintain stability of the anterior tibia bow (Figure 8). Another option could have been osteotomy and revision of internal fixation at the site of the deformity, yet we were concerned about producing a pseudarthrosis.

**Figure 8.** Her rod and screw were removed, and the tibia was protected by inserting flexible nails.

At 9 years of age, and at a routine follow-up appointment, our patient complained of pain in her right leg, difficulty ambulating due to retained hardware. The family was very concerned her tibia would break at any moment despite solid bone on x-ray and with bridging nails (Figure 9).
At this appointment, the family requested that a below-knee amputation be performed. Our patient also had a history of adrenal insufficiency (which may have impacted her osteoporosis and tibial bowing and later fractures). She had a pilocytic astrocytoma that had required multiple surgical procedures with hydrocephalus, VP shunt, and progressive developmental delay. Our patient’s family was further overwhelmed with health issues, as her mother was dealing with her own battle with metastatic breast cancer.

Decision Point #3—Expert Panel

**JL:** Professor Shah and Dr. Iobst, what are the key problems at this point?

**HS and CI:**
- Pain at the tibia with weight-bearing
- Procurvatum deformity of the upper third-middle third junction and middle third-lower third junction
- Valgus deformity of the tibia
- Valgus deformity of the ankle
- Flexible rod with deformity
- Fibular pseudarthrosis
- Poor bone quality
- Leg length discrepancy

**JL:** What are your concerns for this patient?

**CI:** Family has multiple social stresses and limited capacity for complicated post-surgical recovery protocols.

**HS:** Pain is the main concern for this patient. Ambulation with the pain is difficult, and revision of the rod would be difficult.

**JL:** If this were your patient, what would you recommend and why?

**HS:** Rule out infection, rule out re-fracture. Complete blood count, ESR, CRP, CT scan, metabolic work up. Bone scan must be done to rule out infection and re-fracture.

Parenteral bisphosphate with protective clamshell orthosis would be recommended to relieve pain (after ruling out infection and re-fracture).

**CI:** This patient has had multiple reconstructive attempts to rescue this limb without complete success. If the family is requesting amputation, I would agree to this option given their social situation and her young age.

Continued Treatment for Case 3

In retrospect, we should not have been surprised by the burden her leg and the five previous operations had on her and her family. At 9 years and 2 months of age, she underwent below knee amputation and the tibia was capped with a metatarsal head (Figure 10).

Our patient and her parents were very satisfied with her function, and at all follow-up visits for her impending operative scoliosis, they stated they never regretted their decision for amputation. Our patient was participating in her middle school track team, and the family was not dissuaded by the occasional injury from her increased activity (Figure 11).

Presenter Commentary

**JL:** Thank you Dr. Noonan for presenting this case. This case demonstrates the prolonged course that many of these patients have, the multiple decision points along the way, and the overlay of social and medical complexity. In hindsight, if you could treat this patient over again from the beginning, would you do anything differently?
KN: At the time this patient’s surgery was undertaken, the FD Nail and the SLIM™ Nail were not available; thus, I was resigned to use a nail that did not have locking capabilities. The nail migrated after initial placement and required revision. While we modified the rod in our hospital machine shop to make it locking, I wished we had a SLIM™ Nail to avoid this problem. We used the Williams concept of transfixing the hind foot in order to improve distal fixation, and this led to ankle stiffness. I personally follow the philosophy expounded by John Herzenberg who advocates for plating and nailing in order to provide rotational stability. In so doing, you can avoid transfixing the ankle.

Our patient developed anterior bowing throughout her tibia which may be a result of her NF or more likely are- sult of her endocrinopathy and subsequent metabolic bone disease. As a young attending, I followed the dogma of never osteotomizing an intact NF tibia for fear of developing a pseudarthrosis. I therefore tried to span the tibia with nails that could prevent fracture while accommodating the bow. In retrospect, I should have recognized the significance of the bowing and performed corrective osteotomies with an FD nail that spanned the entire tibia and would grow as she did (similar to OI). I would have also bone grafted and plated the osteotomy location(s) with a small plate (à la Herzenberg) to provide rotational control. A good dose of prayer would also be indicated.

JL: Any recommended tips or tricks?

KN: When passing a flexible nail in the tibia, I encountered some resistance and had the “bright idea” of applying a drill to the flexible nail in order to spin it past the tight part in the tibia. Of course, the nail broke. I do not recommend this technique and learned a valuable lesson that titanium nails can fail with torque.

JL: Would you like to emphasize any additional key takeaways?

KN: I have come to appreciate the psychological burden that a diagnosis such as CPT can bring to the family. Every step, jump, or icy sidewalk presents an opportunity for a fracture and need for subsequent surgery and possible morbidity. The effects on the child and the family are also felt in their subconscious, and the fear of the unknown is real. I realize now that patients and families have only so much bandwidth to handle all of the family stresses which we may or may not recognize. For this family, an amputation was an option to remove the uncertainty for an acceptable opportunity to begin a more normal life without her foot.