CASE REPORT

Bilateral patellar tendon-bearing Symes-type prostheses in a severe case of Freeman-Sheldon syndrome in a 21-year-old woman presenting with uncorrectable equinovarus

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SUMMARY

Described is the use of bilateral patellar tendon-bearing Symes-type prostheses in a severe case of Freeman-Sheldon syndrome (FSS) in a 21-year-old woman presenting with uncorrectable equinovarus and multiple surgical sequela of the lower extremities. FSS is considered a type of myopathic distal arthrogryposis; diagnosis requires the following: microstomia, whistling-face appearance, H-shaped chin dimpling, nasolabial folds, and multiple contractures of the hands and feet. Spinal deformities, metabolic and gastroenterological problems, other dysmorphic craniofacial characteristics, and visual and auditory impairments, are frequent findings. We highlight what we believe to be a unique approach to enhancing the patient's ability to ambulate and engage in physical activity, particularly critical determinates of wellness and long-term functional outcome in FSS, when the feet are non-correctable but not amputated. Important implications exist for physiatrists, other rehabilitation providers and surgeons regarding this likely under-diagnosed syndrome.

heavy chain 3 (MYH3; MIM *160720), at 17p-13.1, thought to affect ATP binding to MYH3, cause classic FSS phenotype. Beck *et al* provided further genotype-phenotype correlation evidence.⁵

Described is the use of patellar tendon-bearing (PTB) bilateral Symes-type prostheses in a severe case of FSS in a 21-year-old woman presenting

BACKGROUND

Originally described by Freeman and Sheldon,¹ Freeman-Sheldon syndrome (FSS; MIM #193700) is considered a type of myopathic distal arthrogryposis; diagnosis requires the following: microstomia, whistling-face appearance, H-shaped chin dimpling, nasolabial folds, and multiple contractures of the hands and feet.² Spinal deformities, metabolic and gastroenterological problems, other dysmorphic craniofacial characteristics, and visual and auditory impairments, are frequent findings.^{2,3} Malignant hyperthermia and difficult intubations complicate surgical planning.² Unless there are additional mutations, intelligence is normal; developmental delay results from physical deformities.² Some individuals present with minimal malformation; rarely, patients die during infancy as a result of respiratory complications. Strongest evidence exists for autosomal dominant inheritance, but expression is frequently from new allelic variation. While frequency remains unknown, mostly due to diagnostic uncertainty, there appears to be neither sex nor ethnic preference. Prenatal diagnosis can be achieved via ultrasound or allelic variation analysis.³ Environmental and parental factors are not implicated in the pathogenesis. Toydemir *et al*⁴ showed that allelic variations in embryonic myosin

Figure 1 Patient standing with early trial patellar tendon-bearing (PTB) Symes-type sockets. Notice that while the patient is able to stand pain-free, she does not appear confident due to the bi-valved fabrication of the sockets.

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Figure 2 Craniofacial (A) and hand deformities (B). Notice typical FSS characteristics, including: h-shaped chin dimple, microstomia, nasolabial folds, lack of facial musculature, blepharophimosis, small nose with hypoplastic nasi alae, micrognathia, supraorbital ridge, ulnar deviation, cortical thumbs and camptodactyly.

with uncorrectable equinovarus and multiple surgical sequela of the lower extremities. We highlight what we believe to be a unique approach to enhancing the patient's ability to ambulate and engage in physical activity, particularly critical determinates of wellness and long-term functional outcome in FSS, when the feet are non-correctable but not amputated. Important

Figure 3 Lower extremity photographs (A) and CT (B). In the ankle-foot complex, there is ankylosis of the talocrural joint (left 160°; right 100°) and metatarsals 2–4; weight bearing is on the anterolateral foot (proximal interphalangeal joints). The angle from the origin of the quadriceps to patellar tendon insertion on the tibial tuberosity has approximately a 30° lateral deviation.

Figure 4 Plain radiographic sunrise views of right (A) and left (B) patella.

Figure 5 Lower extremity photographs showing lateral deviation of right (A) and left (B) leg. Notice bilateral patellar subluxation with extension and tibiofemoral joint flexion contracture.

implications exist for physiatrists, other rehabilitation providers and surgeons regarding this likely under-diagnosed syndrome.

CASE PRESENTATION

The patient had an especially severe case of FSS, with past history significant for dysphagia, chronic failure-to-thrive, high intelligence, dysphasia, recurrent acute respiratory complications

Figure 6 Photographs showing passive manual midline bracing of right (A) and left (B) patellae through passive range of motion. Notice that patellae are difficult to hold in midline position, even without joint motion.

and MH-like syndrome, involving surgical (without triggering agents) and non-surgical situations. Beginning at age 2 years, the patient had over 20 surgeries on her feet, knee and craniofacial structures; most were unsuccessful and resulted in deteriorated clinical status. Casting of the feet and bracing of the hands was not followed up by physiotherapy, and corrections were lost. After healing from surgical interventions, several ankle-foot-orthoses were fabricated but were not successful. An early trial socket for a lower extremity prosthesis (figure 1) failed, as it was bi-valved and left the patient's feet unsupported and unprotected.

INVESTIGATIONS

Examination revealed a short, thin female (1.45 m and 30.844 kg, body mass index 14.7 kg/m²) with findings consistent with FSS diagnosis (figure 2), as defined by Stevenson *et al*,² and previously unreported features (McCormick RJ *et al*; Unpublished, 2015). There was significant surgical modification

of the feet, including bilateral talectomies, leaving the feet immobile and heavily scarred with sensory and motor defects, and poorly perfused, as evidenced by essentially non-palpable popliteal and pedal pulses.

The tibiofemoral joints showed bilateral lateral habitual patellar subluxation with extension beyond 120°,⁶ and were laterally deviated (figure 3). Severely hypoplastic lateral epicondyles (figure 4) and a 7–10° lateral deviation of the leg (figure 5), adding lateral force to the patellar tendon, caused patellar malalignment and subluxation. Patellae could be passively maintained in a midline (figure 6) track through full passive range of motion, but not in active range of motion. Straight leg raising was limited bilaterally to about 30–35°, due to tibiofemoral contracture.

DIFFERENTIAL DIAGNOSIS

Differential diagnoses for FSS includes: Sheldon-Hall syndrome, distal arthrogryposis multiplex congenita, Gordon syndrome

Figure 7 Later patellar tendon-bearing (PTB) Symes-type trial sockets, with patient seated in her wheelchair during fitting, showing (A) left and (B) right sockets and (C) standing. Notice these feature typical medial windows and doors, giving the patient good distal weight support.

Figure 8 Final prostheses showing internal padding modifications. Notice various pad types required to accommodate difficult anatomy.

and isolated non-syndromic deformities, but our patient presented with the classic pathognomonic features of FSS of h-shaped chin dimpling, microstomia, whistling face (pursed lips) appearance and prominent nasolabial folds, excluding the possibility of another diagnosis.

TREATMENT

At age 21 years, after using a wheelchair for 15 years, the patient was again fitted for bilateral PTB Symes-type trial sockets, which were not bi-valved and featured medial windows and doors held in place by 1.5" Velcro straps. A large foam base was fabricated to allow the patient to test the sockets indoors (figure 7). In preparation to receiving the trial sockets, a pre-prosthetic lower extremity strength and conditioning programme was developed to assure the patient's ability to ambulate; eventually, concern about patellar subluxation prompted suspension of an otherwise successful rehabilitation period. Exercises were executed on conventional machines in a higher education campus recreational facility and consisted of three sets of 12 repetitions, with 30s between, and focused on quadriceps, hamstrings and hip flexors. After trial socket fabrication, various densities and layers of foam were applied to the interior of the sockets and considerable modification was required due

Figure 9 Patient standing during initial training with final prostheses. Notice patient's confidence and comfort after relatively brief training.

to the difficult anatomical nature of the patient's legs and feet. After these sockets were made comfortable, she did very well with them, and it was decided to create the final prostheses,

Figure 10 Standing with prostheses from front, 8-year follow-up.

Figure 11 Standing with prostheses from right, 8-year follow-up. Prostheses are angled to maintain tibiofemoral joint flexion, preventing patellar subluxation that habitually occurred beyond 120°. Subluxation could also spontaneously occur at any angle.

which would be a bridge to eventual transtibial amputation subsequent to bilateral lateral patellar subluxation correction. The patient was initially fitted with a 23 cm Single-Axis foot connected to her final sockets with a male pyramid fixture, but this was replaced by a Carbon Copy 2 (The Ohio Willow Wood Company, Mt Sterling, Ohio, USA). As with the trial sockets, considerable modifications were required during the initial fitting.

The patient adapted to using the prostheses quickly (figure 8), being stable and comfortable while walking without assistive devices after the initial fitting and training visit of 2 h (figure 9). Except for using parallel bars during the initial training, the patient has never required assistive devices. The patient reported easily continuing her daily routine wearing the prostheses for the

Video 1 Patient rising from a seated position and standing. Observe patient's smooth and confident transition from unsupported sitting with feet together, standing, immediate initiation of gait over uneven surfaces with a step, stopping, and to controlled sitting, without use of arms, and with feet spaced normally.

rest of that day, and reported varied cadence, including light indoor running the following day.

OUTCOME AND FOLLOW-UP

For the first year after receiving the final prostheses, the area of the right foot innervated by the dorsal superficial paroneal nerve was particularly susceptible to pain from pressure that could not be effectively removed, but since there was no skin breakdown in this local area, bupivacaine 0.5% was infiltrated to provide analgesia for 8–12 h while using the prostheses. Gradually, this problem disappeared. For the first 2 years, there was significant difficulty with skin breakdown over most areas on the legs but not the feet, but this was mitigated by changing the liner from stockinette to non-prescription, commercially available nylon compression stockings.

Frequent revisions were required for 3 years, but the patient continued to rehabilitate herself well, achieving an ambulatory level of at least 3 of 4, with an Amputee Mobility Predictor score of 42 of 47 while using both prostheses. Skin breakdown was also essentially eliminated by moisture-wicking, athletic type compression stockings, Nike Elite Compression OTC (Nike USA Inc, Beaverton, Oregon, USA).

Follow-up after 8 years showed sitting, standing (figures 10 and 11) and initiation of gait was rapid and smooth (video 1). Stability continued to be somewhat impaired because of bilateral

Video 2 Patient kicking a ball with both feet. While not shown in the video, the patient did not use her arms for balance. As can also be seen, the prosthetic feet probably need to be adjusted for more eversion, which would have improved the patient's kick.

lateral patellar subluxations, and tibiofemoral joint flexion was limited by the posterior socket rim. The patient was able to kick a ball unsupported, with either foot (video 2), and to comfortably reach out and grasp objects (video 3). While ambulating with bilateral PTB Symes-type prosthetics, there was more trunk motion than expected due to the need to limit tibiofemoral

Video 3 Patient walking and retrieving an object. The patient remains very stable while reaching to retrieve an object. Though performed for demonstration, she is capable of reaching to retrieve large and heavy objects. In university, she carried a backpack weighing 13–18 kg plus a laptop case weighing at least another 5–7 kg. We did not have her retrieve an object from the floor due to her patellar subluxation and spinal deformities.

Video 4 Patient walking a short distance. While walking comfortably, the patient remains very cautious due to risk of patellar subluxation, hence the two-feet turns. The inversion of the prosthetic feet is again visible.

Video 5 Patient walking longer distance over uneven terrain and varied surfaces. Notice that she takes longer strides to compensate for needing to limit the tibiofemoral joint motion.

motion to reduce likelihood of patellar subluxation, and antero-lateral trunk lean was produced by severe scoliosis and lordosis (videos 4 and 5). The patient also extended the hip anterolaterally in swing phase, rather than anteriorly. With concomitant biomechanical limitations, the starting input required was substantially higher than for a physiologically competent Symes or transtibial amputee. When not using the prosthetics, the patient continued to ambulate on her knees, requiring yet higher mechanical input, similar to a transfemoral amputee. While the patient has comfortably worn many types of shoes and sandals, she has best used Nike SFB Field 6" (Nike USA Inc, Beaverton, Oregon, USA), which produces a noticeably more natural gait, better transition and increased stability, even with a unilateral patellar subluxation.

Patient's perspective

- ▶ When I could first stand, and then walk, run and jump without pain or any discomfort on the prototype legs, it was utterly exhilarating! I felt a rush of energy and power. I wanted to just start running and never stop! It was the most wonderful experience, after so many failed attempts to get me walking. I had totally given up on the idea of 'getting better' and didn't believe this latest attempt would be any more successful than anything else in my closet graveyard of failed orthotics.
- ▶ When I got my final prosthetics, I felt like I was on stilts, but then my surgeon, who was there collaborating on their design, pulled my pants legs down to cover the prosthetics, and he asked me to look in the mirror and tell him what I saw. To my astonishment, I saw—for the very first time ever in my life—two nice, normal, straight legs and normal feet. I had seen my new legs as fully part of me—not an extension or device, but a fully incorporated and normal—better—part of my being. After that, I didn't have much trouble getting used to them. I walked with the parallel bars a few times, and then I started walking around the office. I continued the rest of my evening with my new and improved legs, and it was amazing how much easier everything was. I used so much less energy, and it made me feel really great—empowered, independent, able. The total key to walking is to see the legs and feet as mine. As long as I do that, I can do whatever I want without hesitation. If I am timid and don't see them as me, I'll falter.
- ▶ The first feet were good to start with and were nice and stable, but one day, I was late to a very important meeting, and I had to run hard to make it. I had never 'had' to run before, and I felt like I was running through an impossible quagmire while, at the same time, I heard my feet flapping before me with each step. I was pouring sweat with what was very inefficient running, and I felt like my feet were falling apart! I realised I needed new feet that could keep up with me. I am still not real happy with the new feet, but new shoes—Nike SFB—made a big difference, at least as much as changing feet did. Skin breakdown was a big problem for the first 3 years, but I have not had that difficulty with Nike compression running stockings.
- ▶ While my patellae still subluxate regularly, which makes me somewhat unstable, there is nothing better than going from having been in a wheelchair to walking essentially instantly. I feel human and healthy.

Learning points

- ▶ Working with a patient who has not undergone amputation is technically much more challenging and will require more adjustments to the socket after manufacture and subsequently. This need for frequent adjustment over a long duration must be sincerely appreciated by the provider, patient and family, to avoid appearance of failure.
- ▶ The therapeutic goal is to optimise the individual's ability to ambulate and engage in physical activity, not to preserve or reconstruct the patient's biological feet at all costs. This goal may be difficult to envision and accept, especially in the setting of uncorrectable feet that are well vascularised, but it is critical to acknowledge that the patient's ability to ambulate and be physically active will make a larger contribution to the patient's occupational success and good health than preservation of non-functional or marginally functional feet.
- ▶ Patients and parents of young children must understand and accept the therapeutic goal, or all efforts will be lost, even in spite of technical success. Patients and families often believe that the goal is to restore the feet without consideration about what that may mean.
- ▶ Preprosthetic rehabilitation is just as important in patients with congenital foot deformities as reconditioning is for new amputees. De-conditioning is a commonality in both groups.

Since receiving final prostheses, the patient has been wheelchair-free, except for one postoperative and one medial collateral ligament tear recovery period. As the strength and force of spontaneous bilateral patellar subluxations precludes the usefulness of any assistive device, the patient often is accompanied by a physically fit person to assist her in preventing knee injury during patellar subluxation episodes.

DISCUSSION

While the use of PTB Symes-type prostheses in non-amputees may be relevant only in a relatively small patient population, it may successfully act as a bridge to amputation for patients not psychologically or medically prepared for amputation and may represent appropriate destination therapy for patients in whom amputation is not necessary. In a search of PubMed and Google, we did not find any similar reports of using lower extremity prostheses for patients with non-functional feet that had not undergone amputation. While our patient continued to experience patellar subluxations, she was able to remain completely wheelchair-free, and she is very happy with what has been achieved with these prostheses.

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Contributors RJM contributed the syndromic and physiological assessment and gait analysis. MIP, who was the primary originator and drafter of the manuscript, also participated in analysis and interpretation of the findings. RLC advised in the design of the preprosthetic lower extremity strength and conditioning. Rigoberto RT Ramirez, who, sadly, died and did not meet the ICMJE threshold for authorship, provided surgical consultation and advice regarding modifications to the prosthetic design. Mark Gorman, who also did not meet full ICMJE criteria, was the creator of the lower extremity prosthetic devices, reviewed the manuscript, and contributed to the manuscript.

Competing interests None declared.

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