Mobility Challenges and Solutions for Fibrodysplasia Ossificans Progressiva

Charles Levy, MD, Theresa F. Berner, OTL/R, Paul S. Sandhu, MD, Beth McCarty, OTL/R, Nancy L. Denniston, MS, MA


Fibrodysplasia ossificans progressiva (FOP) is a rare genetic disorder characterized by progressive soft tissue ossification. Although signs may be present at birth, the first appearance of ectopic bone typically occurs in early childhood. The primary target is the axial musculature. Eventually ectopic bone also occurs in ligaments, fascia, aponeurosis, tendons, and joint capsules of the appendicular skeleton with a proximal to distal predilection. As the disease advances, mobility becomes restricted, and affected individuals are typically limited to bed or chair by their early 30s. This report describes a 30-year-old woman with advanced FOP. She had a fused spine and a fixed pelvis, with hips and knees locked in flexion and feet in plantarflexion. Her upper limb mobility was similarly restricted. She was not able to stand upright or sit independently.

The modification of a commercially available power wheelchair that allowed the patient to maintain her employment as a preschool teacher and custom shoes are described. Creative physiatric intervention is essential to liberate human potential for people with FOP.

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FIBRODYSPLASIA OSSIFICANS progressiva (FOP) is a rare genetic disorder characterized by progressive soft tissue ossification. Its inheritance is autosomal dominant with full penetrance but variable expression.1,2 Perhaps because of the devastating nature of the disease, few affected individuals have children and therefore new occurrences are usually attributed to new mutations.3-6 The earliest case reports appear in 1692. Prevalence has been estimated at 1 per 1.64 million in the United Kingdom.3 Less than 200 cases worldwide were known as of 1996.5 The cause of FOP is unknown, but is associated with overexpression of bone morphogenetic protein 4 in lymphocytes.6 No effective medical treatment is available. Surgical treatment is nearly always contraindicated, since new heterotopic ossification occurs at the site of any trauma.3 Although there are reports of FOP as early as in utero and as late as early adulthood, the disease typically manifests in the first decade of life. The earliest sign is a short great toe with single phalanx (actually a cartilaginous anlage of the first metatarsal and proximal phalanx) present at birth. Typically between the ages of 2 to 6 years, tender, rubbery, asymmetric, erythematous lumps appear in the paraspinal muscles or limb girdles. These lumps appear over several hours, and remit to variable degrees over the course of days to weeks. Remaining lumps indicate sites of ectopic bone and retard movement corresponding to their location. Involvement of the axial and proximal limb musculature tends to precede that of the distal limb; tongue, extraocular muscles, diaphragm, sphincter muscles, and visceral muscles are usually spared.5,7 At advanced stages of the disease, usually by the third decade, affected individuals are cast in a fixed position, with limited control of only the distal appendages, tongue, face, and eyes.5 Although several promising treatments are poised for clinical trials, no established medical treatment exists. Ineffective treatments have included adrenocorticotrophic hormone, binders of dietary calcium, intravenous infusion of ethylenediaminetetraacetic acid (EDTA), nonsteroidal anti-inflammatory agents, radiotherapy, disodium etidronate, and warfarin.3 Corticosteroids may be useful in acute flares, and there is some indication that iontophoresis with steroids or acetic acid may help restore lost range of motion.10 A study of 25 individuals with FOP ranging from age 5 to 55 years (mean, 22yrs) documented the presence of severe restrictive lung disease.11 Chest expansion was extremely limited (1.9 ± 0.8 inches), suggesting reliance on diaphragmatic breathing. Lung volumes were severely reduced (mean forced vital capacity 44% ± 14% of the predicted value).11 Although individuals with FOP may live into their 70s most die earlier from pulmonary complications.

CASE REPORT

A 30-year-old woman with FOP diagnosed at age 10 presented to our seating and positioning clinic. Progression of the disease had resulted in significant ankylosis. Her entire spine was fused. She was unable to sit because her hips could only flex to 60° and her knees were fixed in 22° of flexion on the right and 30° of flexion on the left. Her ankles were fixed at 55° of plantar flexion on the left and 40° of plantar flexion on the right (table 1). Her gait was slow, effortful, and uncertain. Because her knees and ankles were fixed, she had to rely entirely on the limited excursion of her hips to walk. Because her ankles were fixed in planarflexion, all her body weight was borne by the forefoot alone. Lacking support of the midfoot or hindfoot, she felt vulnerable to falling despite using a cane or a standard walker (fig 1). Painful calluses developed on her metatarsal heads; disproportionate shoe wear was noted at the

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A commercial party with a direct financial interest in the results of the research supporting this article has conferred or will confer a financial benefit upon one or more of the authors.

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Table 1: Available Range of Motion, Lower Limbs

<table>
<thead>
<tr>
<th></th>
<th>Hip Fix</th>
<th>Hip Ext, Pub</th>
<th>Kneecap Fix</th>
<th>Ankle Plantar Fix</th>
</tr>
</thead>
<tbody>
<tr>
<td>Left</td>
<td>40°-60°</td>
<td>Fixed at 30°</td>
<td>35°-45°</td>
<td>40°-45°</td>
</tr>
<tr>
<td>Right</td>
<td>35°-50°</td>
<td>Fixed at 22°</td>
<td>40°-45°</td>
<td>Fixed at 40°</td>
</tr>
</tbody>
</table>

Range of motion is listed in degrees from neutral. For example, left hip flexion is listed as 40°-60°. This means that the patient had a total excursion of 20° starting at 40° of hip flexion and ending at 60° of hip flexion.

Abbreviations: Fix, flexion; Ext Rot, external rotation.

distal sole. Upper limb compromise was also apparent. The right shoulder was severely restricted; the elbow could flex to 60° and extend fully (table 2). She retained near normal right hand function. Left shoulder, elbow, wrist, and intrinsic hand function were severely restricted, and she had had safety problems related to these movement limitations. She was dependent or required maximum assistance for nearly all activities of daily living. Previous interventions had included a home evaluation and modification, a home exercise program, and a weekly ongoing treatment with acetic acid iontophoresis to help restore range of motion to the jaw.

The patient's physical limitations affected almost every aspect of her life. Being limited by endurance, and unable to traverse uneven terrain, the patient spent most of her time indoors. She did not own a wheelchair. When she first presented, she was accompanied by her father. He brought a stool to be set against the wall. Because the patient could neither sit nor stand independently, her father would position the stool so that she could lean against it.

Physiatric Goals

Our physiatric goals were as follows: First, we hoped to increase the patient's comfort and security in ambulation by allowing the ground reaction force to be dispersed by her entire foot, not just the forefoot. Second, we sought a wheelchair that could accommodate her fixed posture, maintain her skin integrity, and ease her transfers. This would reduce the stress on her caregivers of transferring her. Ideally, the wheelchair would allow her to stand upright to gain access to counter tops and ease transfers, and also be capable of tilt to allow access to tables and to relieve pressure. Additionally, a smaller turning radius would allow her to negotiate the narrow confines of the day care center and the church where she was an active member.

Physiatric Interventions

Custom shoes were fabricated with the goal of distributing pressure from the metatarsal heads to the midfoot and hindfoot. Molds were made of the patient's feet. From these, custom compressible foam inserts were constructed. To accommodate the fixed contractures, a foam-in-place custom back and seat were fabricated. Power recline allowed the seat-to-back angle to be increased beyond the standard seating angle of 90° to accept the patient's contours. To exit the chair, the patient would first elevate, then forward-tilt, position her feet at ground level, and then close the seat angle, which gently propelled her out of the chair. To enter the chair, the patient would close the angle slightly so that her back met the seat back initially. Then she would open the angle again to ease the rest of her body into

After an comprehensive search of the commonly available power wheelchairs, we chose to customize a Permobil Chairman Corp. This chair allowed us to build in an anterior tilt of 30°. It also had 8½ inches of seat elevation. To accommodate the fixed contractures, a foam-in-place custom back and seat were fabricated. Power recline allowed the seat-to-back angle to be increased beyond the standard seating angle of 90° to accept the patient's contours. To exit the chair, the patient would first elevate, then forward-tilt, position her feet at ground level, and then close the seat angle, which gently propelled her out of the chair. Transferring into the chair, the patient would close the angle slightly so that her back met the seat back initially. Then she would open the angle again to ease the rest of her body into

the chair would then be lowered and the anterior tilt would be reversed until the seat pan returned to a horizontal position. Because of the severe restriction of motion in her knees, elevating legrests would be necessary to support her calves. The intended use of this chair would be twofold. First, it would allow access to the world outside the home. Second, because the patient could not transfer or rise independently, the proposed power chair would provide independent mobility within the home. Further, it would act as a comfortable chair from which she could engage in ordinary activities such as watching television, eating dinner, or conversing with family or friends.

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Table 2: Available Range of Motion, Upper Limbs

<table>
<thead>
<tr>
<th>Range of Motion</th>
<th>Left</th>
<th>Right</th>
</tr>
</thead>
<tbody>
<tr>
<td>Shoulder Abd</td>
<td>Fixed at 15°</td>
<td>20°-60°</td>
</tr>
<tr>
<td>Shoulder Fix</td>
<td>Fixed at 40°</td>
<td>15°-30°</td>
</tr>
<tr>
<td>Elbow Fix</td>
<td>10°-40°</td>
<td>0°-60°</td>
</tr>
<tr>
<td>Wrist Fix</td>
<td>10°-40°</td>
<td>0°-80°</td>
</tr>
<tr>
<td>Supination</td>
<td>Fixed at 0°</td>
<td>0°-60°</td>
</tr>
<tr>
<td>Pronation</td>
<td>Fixed at 0°</td>
<td>0°-80°</td>
</tr>
</tbody>
</table>

Range of motion is listed in degrees from neutral. For example, right shoulder abduction is listed as 20°-60°. This means the patient had a total excursion of 40° starting at 20° of abduction and ending at 60° of abduction.

Abbreviations: Abd, abduction; Fix, flexion.

the chair (fig 4). Another attractive feature of this chair was front wheel drive which allowed a turning radius of 26 inches when positioned upright. A comparable turning radius in a conventional rear wheel driven power chair is 36 inches.

**Rehabilitation Intervention Outcome**

The custom shoes were well accepted. The patient reported increased comfort and a reduction of calluses over the metatarsal heads. The patient consented to be tested in our gait laboratory. Foot plate studies showed a reduction in peak pressure from 43N/cm² to 35N/cm². Although there is no standard for the maximum acceptable pressure for FOP, increased foot pressures and callous formation are well-accepted risk factors for ulceration in the foot of a patient with diabetes. It is also worth noting that initial contact was made much closer to the heel with a progression towards the toes at lift off, and that her spontaneous walking speed improved from .18 to .19m/sec, which represented a 6% improvement.

The chair was likewise well accepted. The patient's father, who previously had to perform a dead lift dependent transfer, attributed his lessened chronic low back pain to the fact that he now had to provide only contact guard assistance to aid in the transfer of his daughter (fig 4). The patient volunteered that she felt less vulnerable to feelings of depression; she has maintained her occupation as a day care instructor. She reported less fatigue, and has ventured out in public to places such as restaurants, which had been off limits before she had the chair. The patient has returned to her church where she sings with the choir and is a lector.

**DISCUSSION**

FOP is a devastating disease. As heterotopic ossification inexorably advances, mobility becomes increasingly restricted. It is easy to imagine how this results in increased stress on the affected individuals and their caregivers. Since this case report was originally prepared, the primary author has evaluated 20 individuals with FOP at patient-oriented FOP conferences. He also has made himself available to individuals with FOP when he travels to professional meetings. All of those with advanced disease except one were fixed in a semi-upright posture. All of these individuals need complex power wheelchairs not only to provide mobility, but also to aid in positioning and transfers. The features of tilt and recline as found on most commercially available power wheelchairs were designed to aid in pressure relief. These chairs tilt posteriorly, but are not easily modified to tilt anteriorly. To effectively tilt anteriorly, the chair must also elevate. Otherwise the foot rest will strike the ground after an excursion of just a few degrees. The combination of anterior tilt and seat elevation allows individuals such as the patient in this report to assume an upright position. Several chairs are available with a power standing capability, but reach the standing position by rising from a seated position. Individuals with advanced FOP lack the pelvic mobility demanded by these chairs.

The combination of custom seating in a power chair with the capacity to elevate, tilt anteriorly and posteriorly, and recline has applications beyond FOP. Others with extensive contractures or severe joint pain, such as those with arthrogryposis, advanced ankylosing spondylitis, or advanced rheumatoid arthritis could benefit from mobility systems such as the one described.

Custom shoes can distribute pressure throughout the plantar surface, thereby decreasing peak pressures and encouraging tibial advancement over the foot in patients with severe fixed ankle equinus. The increase in gait velocity, along with the patient's report that walking took less effort, suggests that proper custom shoes promote a more energy efficient pattern of ambulation. Therefore, creative physiatric intervention can provide equipment that is essential to liberate human potential for those struggling with this rare but crippling disorder.
Fig 4. The patient rises from a seated position, and exits her wheelchair. (A) The patient directs power chair from a "sitting" position. (B, C) The patient elevates and forward tilts to assume an upright posture. (D) The patient is lowered slightly so her footplates reach the ground. (E) The patient is gently pushed forward by the seat back as the seat-to-back angle closes. (F) The patient's assistant applies minimum force to facilitate the transfer.
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References

Supplier
a. Permobil, 6 B Gill Street, Woburn MA 01801.