

Rehabilitation for Individuals With Fibrodysplasia Ossificans Progressiva

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Abstract

Fibrodysplasia ossificans progressiva (FOP) is a rare genetic disorder characterized by skeletal malformations and progressive heterotopic ossification of soft tissue. As heterotopic bone accumulates, range of motion is progressively lost and leads, eventually, to near-complete immobility. Proper rehabilitation helps to preserve health and function despite great impairment. Present and future rehabilitation approaches to enhance activities of daily living, mobility, education, vocation, and sexuality are discussed.

Key Words: Fibrodysplasia ossificans progressiva (FOP); rehabilitation; iontophoresis; immobility; sexuality; activities of daily living.

Fibrodysplasia Ossificans Progressiva: The Nature of the Problem

Fibrodysplasia ossificans progressiva (FOP) is an exceedingly rare and devastating disorder of progressive soft-tissue ossification (1,2). On a molecular level, the disorder can be traced to the dysregulation of a potent bone-inducing morphogen (bone morphogenetic protein 4) (3). Congenital malformation of the great toe is present in nearly all patients with FOP. FOP usually manifests in the first decade of life, typically between the ages of 2 and 6 yr. Tender, rubbery, asymmetric, erythematous lumps appear over several hours in the paraspinal

muscles or limb girdles. Although some of these masses may remit, most will mature to form true heterotopic bone. This process occurs spontaneously, but may also be triggered by minor physical trauma. Gradually, heterotopic ossification (HO) accumulates in the aponeurosis, fascia, tendons, ligaments, and connective tissues of voluntary muscles, laying down what has been described as a second skeleton.

The transformation/invasion of normal tissue to bone occurs in a cranial-to-caudal, proximal-to-distal, and axial-to-appendicular skeletal pattern (4). Cohen et al. surveyed 44 individuals with FOP and found that the initial manifestations occurred at a mean age of 5 yr and affected the neck, spine, and shoulder. The hip and elbow typically became involved during the 12th year, whereas the jaw, wrist, and ankle were affected between the 15th and 18th years of life. Spinal deformity is also quite common (5). Shah et al. noted that 26 of 40 (65%) individuals with FOP developed scoliosis, and of these, 23 (88%) were “unbalanced

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c-shaped curves.” It is unclear from the report how many were kyphotic, but kyphosis and hip and pelvic rotational deformities and obliquities are common in the authors’ experience. Interestingly, once the body stops growing in the late teens or early 20s, the incidence of spontaneous HO quiets down and becomes less common. Unfortunately, trauma remains a potent trigger throughout life, and may have disastrous functional consequences.

FOP: The Magnitude of the Problem

Statistics fail to convey the true magnitude of the difficulties facing individuals and families affected by FOP. Although there is some variation in expression of the disorder, the formation of heterotopic bone is brutal in its progressive assault during the growing years. Each incident of soft-tissue ossification carries the potential to restrict range of motion in the region where the bone has formed. In small and occasionally larger increments, affected individuals will lose range of motion in the neck, shoulders, spine, hips, elbows, knees, ankles wrists, jaws, and, in some cases, the distal appendages. The diaphragm, heart, tongue, extraocular, and smooth muscles are spared. Thus, by the third decade, barring new trauma, the ravages of FOP will generally be known for each individual. Some will have been lucky and will retain enough hip and lower limb movement to ambulate, perhaps with the assistance of a straight cane, and perhaps will be able accomplish some form of sitting and transferring. However, many affected individuals will find themselves locked in unpredictable postures, typically caught in an asymmetrical pose somewhere between sitting and standing.

The Impact of FOP on Daily Living

In individuals with advanced FOP, almost all aspects of life are impacted, and therefore must be addressed to maximize the individual’s opportunity to live well and fully (6). Issues include the obvious: dressing, grooming, bathing, and mobility, as well as activities that are more often overlooked: sexuality, affect, education, and vocation.

To make matters worse, although steroids and anti-inflammatory medicines may be useful during flare-ups, there remains no effective medical treatment to

halt the progression of FOP bone formation. Surgery is almost always contraindicated because it triggers further episodes of ossification.

Rehabilitation Strategies

Rehabilitation strategies and interventions can be separated into those that are restorative and those that are compensatory. Restorative rehabilitation allows the recovery of skills and abilities lost through injury, disuse, or a disease process. An example of a restorative rehabilitation might be rehabilitation by running and turning in an athlete following a ligament tear or strain. The athlete might be placed on a progressive program of strengthening, flexibility, kinesthetic awareness, and graded return to activity, with the goal of resuming running using the same underlying musculoskeletal structures and neural programming as before his or her injury. Compensatory strategies involve the substitution of new strategies and structures to carry out functions that can no longer be performed using the same neural or musculoskeletal substrates. An example of this would be teaching a person with poststroke hemiplegia how to dress with one hand or how to walk with a quad cane and an ankle-foot orthosis.

In FOP, almost all strategies and interventions are compensatory, many relying on equipment to help preserve and enhance function. Interventions for those with FOP must be tailored to the physical, emotional, and cognitive limitations, abilities, and aspirations of the individuals, as well as the human, financial, and technological assistance available. The construction of the optimal rehabilitation plan often involves the following professionals: physiatrists, occupational and physical therapists, speech therapists, dentists, wheelchair vendors and manufacturers’ representatives, orthotists, psychologists, and vocational rehabilitationists. In addition to professional help, affected individuals and their caregivers can gain a great deal of comfort and practical advice from other families. The International FOP Association (IFOPA), a nonprofit support organization, is a particularly valuable resource. The IFOPA hosts international symposia where those with FOP can meet one another. Additionally, the IFOPA hosts a website, www.ifopa.org, and publishes a newsletter. The website contains valuable medical information, as well as guidebooks for families and children.

Rehabilitation Tactics

Dressing

Typically, the shoulders and neck lose mobility early in the course of the disease, impairing the ability to dress both above and below the waist. A variety of solutions may be employed. For those with limited access to the midline, pull-over shirts and blouses with few buttons or that are pre-buttoned can be helpful. Likewise, loose-fitting clothes with elastic waistbands instead of belts can be used. Sock donners (devices where the sock is placed over a cuff attached to a cord), elastic shoelaces, and long-handled shoehorns may be indicated. Velcro closures may be easier to manage than buttons or zippers, although long-handled dressing sticks and zipper aids are alternative solutions.

Toileting

Two common issues regarding toileting in FOP are the issues of postural abnormalities that prevent sitting, and those that prevent self-hygiene. Raised toilet seats are common commercially manufactured items. Some individuals may require raised and custom-angled commodes to allow the posterior to meet the lip of the toilet. Doorways may need to be widened and grab bars may be needed to allow ingress and egress into the bathroom, and to assist positioning on and off the toilet. Regarding hygiene, long-handled sponges or modified reachers may help ensure cleanliness. Bidets should also be considered.

Bathing and Grooming

Similar considerations as toileting can assist bathing and grooming. Roll-in showers without sills, common or custom shower benches or stools, and hand-held spigots may allow the affected individual or a helper to complete bathing. Long-handled sponges and soaps may provide access to hard-to-reach body parts. Long-handled combs and brushes can enable hair care. Strategically placed mirrors will allow inspection of body parts that would otherwise be obscure. Dental care can be particularly challenging because the jaw loses range of motion. Electric toothbrushes and water pics may be indicated.

Feeding and Meal Preparation

A number of adaptations can be made to assist the task of feeding. Many individuals with FOP may not

be able to eat comfortably in a seated posture. If this is the case, a strategically placed stool and an elevated platform placed on the table may be necessary for upright dining. Long-handled eating utensils and straws may help those who lack the range of motion to feed by ordinary means. These utensils are often fabricated by simply attaching a long wooden handle to a fork or spoon. Dycem[®], a non-slip material, may be placed under plates to prevent them from sliding. If the individual has a limited ability to masticate, food may be ground up or pureed. Food preparation may be made more user-friendly with electrical appliances, such as can or jar openers, cutting boards with spikes to hold food while it is prepared or cut, and rotating shelves (such as a “lazy Susan”).

Mobility

At various stages, canes, walkers, wheelchairs, and orthopedic shoes may be necessary. For those who can still stand or walk, a shoe with a custom-molded insole and build-ups (usually in the heel) allow improved stability and greater pressure equalization throughout the foot (6). Canes are available in a variety of conformations and materials. Cane handles with ergonomically designed grips or that are even custom-molded to the hand are preferred to minimize potential trauma to the hand, elbow, and shoulder. Tips are usually made of rubber, but some are designed to flex and absorb shock. Pointed devices that flip down at the cane tip can increase traction in ice. Canes with multiple tips are available as “quad canes” in smaller and larger bases to increase stability, but at the expense of decreased maneuverability and increased weight. Forearm and axillary crutches may improve center-of-gravity control. Padded covers should be placed over the forearm portion to decrease friction and to cover sharp edges. Similar adjustments should be made for the axillary portion of axillary crutches. Walkers generally provide greater stability than canes and crutches, but may be clumsy. They must be adjusted to the user. Wheels of various diameters, either static or swivel, are available, as are seats, baskets, and hand breaks. Walkers can be equipped with platforms and arm troughs to better fit certain skeletal deformities.

Manual wheelchairs are usually of little benefit for independent mobility in FOP because the upper limb ankylosis is likely to preclude wheeling before

the ability to ambulate is lost owing to lower limb impairment. Power wheelchairs can be very helpful but usually must be adjusted and customized to the particular user's posture and range of motion. This usually demands custom-molded seating and adjustments of the footplates, leg rests, seat-to-back angle, and armrests. The power functions of seat elevation and depression, anterior and posterior tilt, and the ability to open and close the seat-to-back angle is often necessary in those with extensive involvement to allow getting into and out of the power chair (5). The joystick should be placed within easy reach of the hand. Lap trays with mounts for laptop computers allow participation in work and school. These trays should be easy to remove or flip out of the way for transfers.

Vocational and Educational Issues

Until a practical and comprehensive treatment is developed for FOP, affected individuals and their caregivers should be prepared to participate in school and work from a fairly sedentary position. For children, the need for normal exploration and physical activity will have to be tempered by the knowledge that accidental trauma may result in increased permanent disability. For example, non-contact, low-impact sports and recreation may be an acceptable compromise between avoidance of danger and full participation. However, there is no set rule. These decisions must be made at the individual and family level. It is sensible to encourage intellectual pursuits and to encourage computer skills. Levy et al. demonstrated that participation in educational conferences for individuals with FOP could be enhanced by broadcasting them over the internet with streaming video (7). Public school systems in the United States must provide each disabled child with an individualized educational plan, and an education in the least restrictive environment. Children are entitled to occupational, physical, and speech therapy, as well as classroom aides, if indicated. Each state is required to offer some sort of vocational rehabilitation to help people with disabilities enter or remain in the workforce. This may be a source of funding for adaptive equipment, such as computers or power chairs, tuition, etc., as well as evaluations and guidance from vocational counselors. It is worthwhile to remember that, despite the limitations imposed by FOP, most affected individuals will

live normal or near-normal life spans. With the proper consideration, accommodation, and equipment, individuals with FOP can participate and contribute meaningfully in work and school.

Hearing

Undiagnosed hearing loss may be mistaken for a learning disorder. Because conductive hearing loss is common in FOP (presumably because of involvement of the inner ear), biannual audiology evaluations are recommended for children with FOP, and annual evaluations are recommended for adults. This is important not only to increase participation, but also for safety reasons. Hearing becomes an especially important avenue to monitor the environment for those who are unable to rotate the head, neck, and spine. Both air-conduction and bone-conduction hearing aids may be helpful. Some units have remote controls, a useful feature for those unable to reach their ears (8).

Transportation

Great care must be taken to transport a person with FOP who must use a power chair. By the time a power chair is necessary, most people with FOP are unable to brace or protect themselves should they start to fall. The result can be catastrophic. Not only is there a high risk for bruises and broken bones, but there also is risk of head trauma and brain injury (9). If the affected person must rely on public transportation, the personnel handling the lift must be well trained and attentive in order to avoid accidental falls or bumps. Persons with advanced disease will have great difficulty driving, although automotive adaptations may allow some to achieve driving independence. A number of strategies are available for modifying vans to accept someone with FOP who must remain in his or her power chair. Ramps and lifts can be installed, roofs can be raised, floors can be lowered, and new controls and motors can be installed to allow the van to "kneel," lowering ground clearance to ease ascent into the van. Many manufacturers offer rebates for van modification to accommodate those with disabilities.

Home Modifications, Aids, and Attendants

Home modifications for those with FOP include external ramps, elimination or minimization of indoor steps, grab bars, widened hallways, and accessible

bathrooms and kitchens. Light switches should be placed where an affected individual can reach them. Environmental control units allow a person with limited mobility to control appliances, doors, televisions, and telephones remotely. Telephones should be situated so that the person with FOP can talk privately. The telephone speaker and microphone may need to be separated, with the speaker and microphone close to the face, while the keypad is placed near the fingers. A headset attached to a standard or cellular telephone may suffice.

Sleep

Restful sleep can be difficult to attain for patients with FOP. For some, particularly those with lesser involvement, bolsters and pillows may suffice. However, many individuals with FOP are unable to turn at night or even to get into bed independently. Tilt table beds allow the bed to rotate from vertical to horizontal. The user approaches the bed when it is vertical, places his or her feet on a platform at the foot of the bed, and the bed then rotates to the standard horizontal position. Specialized surfaces to evenly distribute or redistribute pressure, such as low-air-loss mattresses, help protect and promote skin integrity.

Many individuals with FOP will require the assistance of a caretaker. Family members often find themselves in this role, but in other cases, helpers must be hired. Because of the complex nature of FOP, the risk of pressure ulcers, and the amount of assistance required, a professional, such as a licensed practical nurse, is preferred. This increases the chance of quality care, early recognition of medically urgent situations, and a stable home environment.

Sexuality

Sexuality is an important component of human identity. Physical limitation imposed by FOP may lead to feelings of unattractiveness, isolation, or shame. Individuals with FOP may end up relying on their families for physical assistance during adolescence and beyond, during a time where independence is being forged in normal development. The presence of caregivers may decrease opportunities to experiment with intimacy with appropriate-aged mates, resulting in delayed development. The actual acts of sexual intimacy often require tact and thoughtfulness. Pillows and bolsters may be necessary to support the

unusual and inflexible postures imposed by FOP. Several issues should be considered regarding reproduction. First, limitations in posture and joint mobility may place the woman's or the fetus' health at risk in the case of pregnancy. Second, FOP is an autosomal-dominant disorder with full penetrance but variable expression. Genetic counseling and guidance regarding issues of contraception are warranted for the sexually active or those who are considering such activity.

Aquatic Therapy

Pool therapy offers unique benefits to those with FOP. Buoyancy in water allows individuals to perform active range-of-motion, cardiopulmonary, and resistive exercise in a safe, low-impact environment. Parallel bars can be submerged to allow ambulation training. Therapists or caregivers can deliver stretching and resistive exercises with less biomechanical stress than might ordinarily be encountered on a mat. Warm water can facilitate pain relief and relaxation, and may be able to reach skin folds difficult to access otherwise. Modified lifts, elevators, or ramps may be necessary for entry and exit.

Iontophoresis

Iontophoresis involves the introduction of topically applied, physiologically active ions through the epidermis using continuous direct current. A case study reports the successful use of iontophoresis to deliver a 2% acetic acid solution to a young athlete who developed traumatic myositis ossificans (10). Steroids are often delivered by iontophoresis. Anecdotal reports exist of iontophoresis and range-of-motion exercises in the restoration of jaw motion in FOP. If verified, an iontophoresis regimen could be considered restorative rehabilitation for FOP.

The Future of Rehabilitation in FOP

New technologies are being developed that can augment the armamentarium of solutions for those with FOP and other disabling conditions. One emerging therapy is the "smart house," such as that under development at the University of Florida (11). Smart homes use sophisticated tracking and computer technology to assist those with cognitive and physical disabilities. They can be programmed to respond to simple voice or remote control commands from within or outside the home to operate

door locks, lights, small appliances, thermostats, and windows/curtains. Some household functions can be automated. On-off cycles for heating/air conditioning units, security systems, lights, televisions/stereos, lawn sprinklers, music, and lighting can have established weekday, weekend, and vacation modes that can be revised remotely via a telephone call. Vital signs can be recorded to monitor health status. Behaviors, such as trips to the bathroom, visits to the kitchen/refrigerator, time spent in bed (sleeping, tossing and turning), and time spent exercising or sitting in the living room, can be monitored and recorded and analyzed. If there is a significant deviation from that behavior, the smart house can check with the resident or alert a family member that there may be a problem. An alert can be issued if the resident falls, notifying the caregiver or emergency services where the person is located in the house. The smart house detects when the mail has been delivered, when someone is at the front door, when the stove has been left on too long, or when the resident has gone without medication or food and water. The smart house can also monitor itself to see if the furnace filter needs to be cleaned, if medicines should be ordered, or if the dishwasher is broken. It can handle such details as preparing grocery lists, ordering and arranging for delivery of groceries or medications, or arranging for someone to make a home repair.

Conclusions

A great amount of effort is being directed to developing treatments for FOP (12). If the process of HO can be halted long enough to permit safe surgery, then true restoration of function can be envisioned. Many new challenges will present when that day comes. How should surgery be staged, and what sites should be approached first? How will muscle and connective tissue that have been immobilized in shortened positions for years respond to new degrees of free movement? What will be the optimal methods

to restore walking, breathing, swallowing, and reaching? These questions will fall squarely in the laps of the rehabilitationists working alongside the molecular medicine specialists and surgeons caring for individuals with FOP.

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