Bound by Bone: A Look at Fibrodysplasia Ossificans Progressiva

An Honors Thesis (HONRS 499)

by

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Abstract

Fibrodysplasia Ossificans Progressiva, also known as FOP, is a devastating and debilitating genetic disease characterized by the formation of heterotopic bone. By exploring the pathophysiology, current treatments, and recent research we take an in-depth look at this rare and unfamiliar disease. Through application of the nursing process, nursing care of patients affected with FOP is discussed in the second half of the paper. Even though this is an incredibly rare disease, awareness regarding FOP needs to be brought to the forefront. Knowledge is power. Only through education can people be armed with the knowledge needed to take proper care of a person affected with Fibrodysplasia Ossificans Progressiva.
Acknowledgements

There are several people whom I would like to recognize for their support in the development of this paper. I would like to thank my advisor, Dr. Renee Twibell, for all of her support during this extensive endeavor. Without her words of wisdom and constant encouragement, this paper would not have developed into what it has today. This topic would still be a simple idea in my head. I would also like to thank my husband, Chris Bassler, who provided me the emotional support and encouragement needed during those nights I stayed awake preoccupied with this project. He also assisted in the design of the cover. Thanks also goes to all those suffering or affected by this devastating disease. Without them, there would be no paper about Fibrodysplasia Ossificans Progressiva. Lastly, I would like to thank all the scientists and researchers involved in finding a treatment and cure for FOP. It is because of their dedication that gives hope to the hundreds affected with FOP.
Fibrodysplasia Ossificans Progressiva

A young couple take their three year-old son into the doctor's office. They are concerned about the painful, warm, red mass that has appeared on his back. The mass first appeared approximately twelve hours ago and was originally dime-sized. It is now the size of a grapefruit.

A thirty year-old female wheels around the mall in her motorized wheelchair. Two years earlier she became confined to her wheelchair after tripping in her house. Both her hips are fused and permanently immobile. While her right arm is free to move, her left arm is frozen in a right angle to her side.

What do these two people have in common? They both are afflicted with a disease known as Fibrodysplasia Ossificans Progressiva, or FOP. FOP is an extremely rare genetic disorder characterized by excessive bone formation. Until recently, there has not been any research performed on this extraordinary and devastating disease.

Overview

Fibrodysplasia Ossificans Progressiva, formerly known as Myositis Ossificans Progressiva, is a rare genetic disorder characterized by the formation of heterotopic bone. FOP literally means "soft, connective tissue that progressively turns to bone." (Wagman, Kantanie, & Kaplan, 2001)

Pathophysiology

In FOP, heterotopic ossification occurs. This means that bone forms where bone does not belong, such as in the elbow or hip joints. This bone is "normal" bone; the abnormality is not the bone but its timing and location of formation. Microscopically, this bone is identical to "normal" bone. On an x-ray, the bone appears as "normal" bone. If this bone breaks, it repairs itself as "normal" bone does. Just like "normal" bone, this bone even contains marrow. (Maeder, 1998) As this extra bone continues to form, it eventually makes a "second skeleton."

The formation of the bone is unpredictable in its rate and severity. It affects each individual differently. The one common denominator between FOP patients is
the predictable pattern; the anatomic progression is strikingly similar among each individual. (Treatment Considerations, 2001; Lynch, 1999) Formation of the bone is not random, but mirrors skeletal formation beginning in the womb. The heterotopic ossification process follows the same pattern as in embryonic skeletal development: cephalocaudal and proximal-distal. The ossification begins with the connective tissue in the neck and spine. It next affects the shoulders followed by the hips and elbows. Knees and wrists are affected next then the ankles. Lastly, the jaw becomes ossified. (Maeder, 1998) Fortunately, the muscles of the diaphragm and heart are not affected. Because of this similar pattern of bone formation, the body is in essence not just forming extra bone but rather a “second skeleton.” This “second skeleton” eventually binds the individual in bone, leading to ankylosis and complete immobility. (Treatment Considerations, 2001)

As mentioned previously, heterotopic ossification follows a specific pattern but the rate at which it progresses and its severity for each individual differ. A flare-up is when FOP exacerbates and usually involves the presence of a lesion. This is when ossification of the bone actively occurs. These flare-ups are sporadic and unpredictable. A person may go months or years without experiencing a flare-up. When an individual experiences a flare-up, it is impossible to predict the duration or severity. Flare-ups usually occur as a result of some type of trauma to the body. The trauma can be as severe as a fall or as minor as an intramuscular injection during a routine vaccination. In fibrodysplasia ossificans progressiva, any type of trauma to the body, no matter how minor, can trigger a sudden flare-up. Flare-ups can also occur spontaneously and at random with no precipitating factor. A person can go to bed one night able to walk and wake up the next morning permanently bound to a wheelchair. At this point in time, it is unclear why this disease is active at times and why it lies dormant at other times. (Treatment Considerations, 2001; Wagman, Kantanie, & Kaplan, 2001)

It is still a mystery as to why this bone decides to for, although through
research there have been a number of discoveries that may eventually lead to a definitive answer as to why this bone forms. In the body, bone formation occurs in one of two ways: intramembranous formation or endochondral formation. In intramembranous formation bone cells called osteoblasts secrete a bony substance on top of the bone. This is similar to bricklayers stacking bricks to build a wall. This type of formation is what causes the shaft of the long bones to widen and the top of the skull to thicken. Endochondral formation is how the bones grow longer and how fractures heal. In this process, the body forms a cartilage model. Consequently, it is infiltrated, reabsorbed, and replaced by bone. An example of this bone formation occurs when the fetal skeleton develops. In the disease process of FOP, heterotopic bone forms using the endochondral pathway. (Maeder, 1998)

Why does heterotopic bone form in patients with FOP? Researchers have stumbled across a few reasons why the progressive growth of bone, known as heterotopic ossification, occurs in FOP patients. One reason involves bone-producing proteins called bone morphogenetic proteins, or BMPs. In FOP, researchers specifically look at BMP4, a master protein involved in the formation of the skeleton. (Wagman, Kantanie, & Kaplan, 2001) The “normal” job of BMP is to create bone during embryonic development. (Lynch, 1999) In patients with FOP, BMP production is excessive. The overproduction of BMP, specifically BMP4, causes heterotopic ossification. (Tenth Annual, 2001; Lynch, 1999) It has been discovered that the lymphocytes in FOP patients carry the bone-producing protein BMP4. Normally this protein is not found in lymphocytes. The presence of BMP4 in the lymphocytes highly suggests why bone formation occurs easily after an injury. When an injury or trauma to an area occurs, lymphocytes “swarm to the site” with their doses of BMP4 causing ossification at the site. (Maeder, 1998) It is this inappropriate expression of BMP4 that causes heterotopic ossification. (Tenth Annual, 2001) Under “normal” conditions, there are “safety switches” to tell the body to stop producing BMP4. So, if a cell where even to begin producing the bone-producing protein, the
body would halt the process of bone formation before it occurs. (Tenth Annual, 2001) Cells of an FOP patient are unable to react appropriately to the increased presence of BMP; there is a disturbance between BMP4 and its antagonists. (Tenth Annual, 2001)

Researchers have recently discovered Noggin, a protein involved in the regulation of the concentrations of BMP4 available to the body. (Tenth Annual, 2001) Initially the discovery of Noggin held much promise in finding the real cause of heterotopic ossification. Noggin works by actively binding to BMP4 and inactivating it. (Tenth Annual, 2001) It was hypothesized that there must be a malfunction in the expression of this protein. Later, it was discovered that there were no mutations found in the Noggin gene. (Tenth Annual, 2001) Even though this was a disappointment, Noggin still holds promise in helping to find a cure for FOP. In essence, FOP patients have lymphocytes that over express BMP4 while simultaneously under expressing the proteins that block and suppress BMP4. (Treatment Considerations, 2001)

Angiogenesis plays an important role in the formation of heterotopic bone. Scientists have recently discovered the overproduction of basic fibroblast growth factor during FOP flare-ups. It is circulated in the blood and excreted in the urine. This protein stimulates the production of new blood vessels (angiogenesis) during flare-ups. (Tenth Annual, 2001; Wagman, Kantanie, & Kaplan, 2001) One important factor of bone formation is adequate blood supply. Identifying the presence of this protein during flare-ups holds promising treatments in the future. Current treatment research is now focusing on this aspect of FOP.

Another important factor to consider with FOP is the presence of mast cells. Mast cells are intimately involved during the flare-ups associated with FOP. (Tenth Annual, 2001) It has been found that there is an abundance of mast cells present at every stage of the disease process. (Treatment Considerations, 2001) At this time, it is unknown of the exact part mast cells play in the heterotopic ossification process. It is believed that they play a very important role is the disease process of FOP.
Scientists hypothesize a possible autoimmune component to the disease. Researchers are beginning to explore the possibilities of the possible role of the immune system in FOP. (Tenth Annual, 2001)

**Etiology**

FOP is an autosomal dominant genetic disorder. This means that a person who carries the FOP gene will express the gene; the person will have FOP. (Wagman, Kantanie, & Kaplan, 2001) In 1998, it was identified that the location of the gene was on the long arm of chromosome four. This discovery will make it easier to pinpoint the exact location of the FOP gene someday down the road. (Tenth Annual, 2001) A person who carries the gene for FOP has a 50% chance of passing the trait on to his/her offspring. (Wagman, Kantanie, & Kaplan, 2001) Presently, there are three families in the world known to have the gene and passed it on to offspring. (Maeder, 1998) Almost all known cases of FOP have resulted from the spontaneous mutation in the gene. (Wagman, Kantanie, & Kaplan, 2001; Maeder, 1998)

**Incidence/Prevalence**

Fibrodysplasia Ossificans Progressiva affects an estimated 1 in 2 million, or 2,500, people worldwide. (Wagman, Kantanie, & Kaplan, 2001; Lynch, 1999) To date, researchers are aware of fewer than two hundred cases throughout the world. (Wagman, Kantanie, & Kaplan, 2001) It affects both men and women, both young and old. Symptoms of FOP usually appear within the first or second decade of life; most people know that they are affected with FOP by the age of ten. (Wagman, Kantanie, & Kaplan, 2001)

**Assessment**

Physically, there are two similar features of FOP patients: the malformation of the great toes and heterotopic ossification later in life. (Wagman, Kantanie, & Kaplan, 2001) The malformation of the great toes acts as a very early warning sign of FOP prior to the onset of heterotopic ossification. (Wagman, Kantanie, & Kaplan, 2001) At birth, children appear "normal" except for a congenital malformation of
the great toes. The great toes are generally small and misshapen. The toe may be
turned inward and the joint is often missing. (Lynch, 1999) There is a very small
percentage of FOP patients who do not have the congenital malformation of the great
toes at birth. These children demonstrated arthritic-like changes in their toes by the
age of ten. (Wagman, Kantanie, & Kaplan, 2001)

The heterotopic ossification process in these patients often begins with a
“flare-up.” Flare-ups typically mark the beginning of the bone formation process
and may last as long as 6-8 weeks until the bone is completely formed. (Wagman,
Kantanie, & Kaplan, 2001) These flare-ups are often painful; the pain usually
subsides once it is over. (Wagman, Kantanie, & Kaplan, 2001) FOP is not chronically
painful. (Wagman, Kantanie, & Kaplan, 2001) The formation of lesions usually
characterize the flare-up process. In a flare-up, the affected area becomes swollen,
red, hot, and painful. Within days or weeks, this area turns to bone. (Wagman,
Kantanie, & Kaplan, 2001; Maeder, 19984) This is the process known as heterotopic
ossification. As mentioned previously, these lesions are painful. After the lesion has
developed into bone, they no longer hurt although they may still be a source of
discomfort to the person due to excessive pressure in the area of bone formation.
(Wagman, Kantanie, & Kaplan, 2001)

An initial flare-up is often misdiagnosed as a tumor. The lesions, in their
intermediate stage, are microscopically indistinguishable from a type of tumor called
juvenile fibromatosis. (Treatment Considerations, 2001) This can lead to devastating
effects. Attempting to remove the lesion or bone results in even more aggressive
bone formation. (Wagman, Kantanie, & Kaplan, 2001)

Flare-ups tend to differ slightly in adults and in children. While children tend
to have nodular flare-ups characterized by distinct lesions, adults tend to have a more
“sheet-like” flare-up. In other words, adults tend to have swelling occurring in the
entire limb instead of in the form of a lesion. Even though there is a tendency for
one type of flare-up to occur during a certain age, either form of a flare-up can
occur at any age. (Wagman, Kantanie, & Kaplan, 2001) Eventually, as a result of multiple flare-ups, ankylosis occurs rendering movement impossible. (Treatment Considerations, 2001; Lynch, 1999) People affected with FOP may end up fused in a standing position or in a twisted sitting position. (Maeder, 1998) There is no way of knowing how a person will be "locked" for an eternity. Most FOP patients are confined to a wheelchair by the time they are in their early twenties. (Treatment Considerations, 2001)

FOP is not a fatal disease in itself, but it can cause fatal complications for those affected with the disease. One life threatening complication of FOP is the effects of heterotopic ossification on the chest wall. The chest wall eventually has grids of bone that can restrict breathing. The severe restrictions on the chest wall places the patients at an increased risk for cardiopulmonary problems. Cardiopulmonary complications are believed to play a major role in the shortened lifespan of some FOP patients. Another life threatening complication occurs when the jaw fuses. This can result in severe malnutrition and in some cases starvation. (Treatment Considerations, 2001; Wagman, Kantanie, & Kaplan, 2001; Lynch, 1999; Maeder, 1998) Despite these horrific complications, there are muscles that are fortunately spared from heterotopic ossification. This includes the diaphragm, the heart, the tongue, the eyes, and the face. (Wagman, Kantanie, & Kaplan, 2001) Another common feature of FOP among patients is conductive hearing impairment. At this point in time it is a poorly-understood problem associated with the disease. (Treatment Considerations, 2001)

"One feature that marks victims of FOP, one that strikes everyone who meets them, is their ability to take the unbearable in stride." (Maeder, 1998) FOP is a disease that obviously can have some psychosocial ramifications. All people adapt differently. Children who are affected by FOP themselves tend to cope better than their parents. (Wagman, Kantanie, & Kaplan, 2001) It has also been noted that the earlier a child is affected with the physical symptoms and limitations of FOP, the
easier it is for the child to cope psychologically with the physical restrictions imposed on them. (Wagman, Kantanie, & Kaplan, 2001) Resources that are useful for patients, parents, family, and friends include: professional guidance, family counseling, and meeting/writing with other members of the FOP community. (Wagman, Kantanie, & Kaplan, 2001)

There is no one definitive laboratory test that aides in the diagnosis of FOP. Through research, it has been identified that an excessive amount of mast cells are present during each stage of lesional development during flare-ups. To see the mast cells under a microscope, a special dye must be used to identify them. (Tenth Annual, 2001) In the preliminary stages of research, a muscle biopsy was taken from a patient affected with FOP to investigate any abnormalities in the muscle tissue. Much to the disbelief of researchers, the muscle biopsy indicated 100% normal muscle tissue. Unfortunately, a month after the biopsy, bone regrew in its place. (Skeleton Key, 1999)

Radiographic x-rays can be used as a non invasive way to show the presence of a lesion. In early lesion development, the x-ray indicates only soft tissue swelling. Eventually, in the later stages of lesion development, the x-ray shows the new “normal” bone that has formed. Bone scans are another non-invasive radiographic technique used in FOP patients. These scans can show us formation of new bone early on in the course of a flare-up. (Wagman, Kantanie, & Kaplan, 2001)

Treatment

Presently, there is no prevention or treatment available for those affected by FOP; it is an incurable disease. The present treatments can be divided into two separate categories: symptom-modifying and disease-modifying. At this time, there are no disease-modifying therapies. (Wagman, Kantanie, & Kaplan, 2001) However, there are ways to manage the symptoms of flare-ups and preventative measures to take to reduce the risk of complications and/or flare-ups associated with the disease. Medications for FOP are used to minimize the pain associated with flare-ups.
Current medications used for FOP can be classified into three classes. (Treatment Considerations, 2001) Class I medications are medications that have been used to treat the symptoms of an acute flare-up. These medications have anecdotal reports with favorable clinical trials. They also have minimal side effects. Examples of class I medications include: corticosteroids, non-steroidal anti-inflammatory drugs (NSAIDs), and cyclo-oxygenase 2 inhibitors (Cox-2 inhibitors). Short-term use of prednisone has shown to be effective in the management of symptoms during an acute flare-up. Prednisone decreases lymphocyte recruitment and tissue infiltration. It is also a very potent anti-inflammatory medication. In order for prednisone to be effective, it must be started within 24 hours of the onset of the flare-up. The first 24 hours corresponds with the earliest phase of acute infiltration of lymphocytes into the skeletal muscle. Prednisone must not be used for longer than four days. Prolonged use of steroids has not proven to be of any benefit and actually may help accelerate heterotopic ossification. Although prolonged use is contraindicated, there is one indication for long-term use of steroids. If the flare-up involves the submandibular region long-term therapy can be used. These flare-ups can be life-threatening as it can interfere with breathing and swallowing. The use of steroids has not been effective in treating flare-ups involving the trunk of the body. (Treatment Considerations, 2001)

Non-steroidal anti-inflammatory drugs, also known as NSAIDs, are commonly used in the management of symptoms during acute flare-ups. NSAIDs typically used include ibuprofen (Advil or Motrin) and indomethacin (Indocin). Both medications are helpful with the symptomatic management of FOP because of their anti-inflammatory and anti-angiogenic properties. (Treatment Considerations, 2001)

Cyclo-oxygenase 2 (Cox-2) inhibitors include the medications celecoxib (Celebrex) and rofecoxib (Vioxx). These two medications have properties much like the NSAIDs, except they have fewer side effects. One problem with the Cox-2 inhibitors is that they should only be used in patients older than sixteen years of age.
Class II medications are medications that theoretically apply to FOP. These medications have been approved for treatment in other disorders and have few side effects. The types of medications in this category include leukotriene inhibitors and mast cell stabilizers. One type of leukotriene inhibitor currently used for FOP is montelukast (Singulair). This medication blocks inflammatory mediators in the inflammatory process. It also complements the action of Cox-2 inhibitors. Cromolyn (Gastrocrom) is the mast cell stabilizer currently used for FOP. Mast cell stabilizers reduce mast cell degranulation. One disadvantage of this medication is that it is poorly absorbed in the gastrointestinal tract. (Treatment Considerations, 2001)

Class III medications are investigational drugs. These include the following medications: Thalidomide, Squalamine, and Noggin. Thalidomide has anti-angiogenic properties and is also an immuno-modulator. Squalamine also possess anti-angiogenic properties. Noggin works by blocking the action of BMP4. (Treatment Considerations, 2001)

Iontophoresis is a treatment currently being explored in the management of FOP symptoms. It involves using electrodes at a low electrical current to deliver medication deep into muscle tissue. One combination of medication used in this treatment is a steroid called dexamethasone and a local anesthetic called lidocaine. By delivering these medications deep into the muscle tissue, many patients have found relief from the pain associated with flare-ups. There is no evidence at this time that supports the fact that this treatment will change the course of the disease. It does however provide pain relief. (Wagman, Kantanie, & Kaplan, 2001)

Many people wonder if the extra bone that forms can be removed surgically. The answer is yes, it can, but with severe consequences. This is traditionally not an option for FOP patients. By removing the extra bone with surgery, more trauma is caused to the area and more extensive bone growth results. (Wagman, Kantanie, & Kaplan, 2001)
Injury prevention plays a big role in the management of FOP. By preventing injury and trauma to the tissues, many flare-ups can be avoided. Intramuscular injections should be avoided. By injecting a needle directly into the muscle, trauma to the tissue results. Even though this is a minor "trauma," in people with FOP it can lead to a painful flare-up. Other types of injections are tolerated fine with FOP patients. People affected with FOP should also receive annual flu shots administered subcutaneously. By receiving a flu shot, the chances of having the flu during flu season are greatly decreased. In patients who have restricted chest wall expansion, a flu shot could save his/her life. Avoiding any type of activity that has a high risk for injury is recommended. Contact sports should not even be considered because of the devastating outcomes that can occur. (Wagman, Kantanie, & Kaplan, 2001)

Unfortunately, physical therapy is contraindicated in people with FOP. It does not help in maintaining joint movement. In fact, extensive physical therapy can actually lead to a painful flare-up. Active range of motion is okay and should be encouraged. (Wagman, Kantanie, & Kaplan, 2001)

Recent Research

Research in the area of FOP is extensive at this time. Many people are putting a lot of time and effort to not only find an effective treatment for FOP but also a cure. Gene therapy and gene correction are an evolving technology. Researchers are considering this as a possible cure for FOP one day. It is too early to tell if this would be an effective treatment someday. There is still a lot of work and research that have to occur in the development of gene therapy in general before applying it specifically to a disease such as FOP. Scientists still feel that one day gene therapy and gene correction with the use of the protein Noggin may provide a viable option. (Treatment Considerations, 2001; Tenth Annual, 2001)

Advances in today's research suggest that the use of stem cells may hold the key to a cure for FOP. This is an area that has not had much physical application in research but theoretically it holds promising results. (Treatment Considerations,
Cyclooxygenase 2 inhibitors are currently used in the symptomatic treatment of FOP during acute flare-ups. Current data suggests that with prostaglandin inhibitors such as Cox-2 inhibitors heterotopic ossification can be prevented. For these prostaglandin inhibitors to be effective in preventing the formation of bone, the medication must be at a certain level in the blood before the onset of a flare-up. This is an area to continue to look into for the prevention of bone formation in FOP patients. (Treatment Considerations, 2001)

Anti-angiogenic agents are medications that inhibit new vessel formation (angiogenesis). Angiogenesis is a major contributor to lesion formation and heterotopic ossification. Researchers believe if they can inhibit the growth of the new blood vessels, then they can stop the process of heterotopic ossification. Theoretically this is true. Presently, there are a number of anti-angiogenic agents being researched for their uses in the FOP disease process. One is called Squalamine. Discovered by accident by Dr. Michael Zasloff in 1992, Squalamine expresses potent anti-angiogenic properties. It is anticipated that this agent will target the early and severe pre-osseous flare-ups. At this point in time there is no absolute guarantee that squalamine will be an effective treatment for FOP, but there is hope. (Treatment Considerations, 2001; Tenth Annual, 2001)

Thalidomide is another anti-angiogenic agent currently under research for FOP because of its angiogenic properties. In Europe in the 1950’s, it was used as a sedative. It was generally tolerated well by patients. Then, in 1961, birth defects were reported. These birth defects were eventually linked to thalidomide. The birth defects, which included various defects of the limbs, were due to the inhibition of angiogenesis during fetal skeletal formation. Thalidomide inhibits fibroblast growth factor which induces the formation of blood vessels. It is also an immune-modulator. This means that it controls reactions of the immune system and it could possibly play a role during the early development of FOP lesions when the lymphocytes attack the
muscle tissue forming bone. In August 1998, phase I of the thalidomide trial began. (Treatment Considerations, 2001; Tenth Annual, 2001) The primary objective of this study was to “determine the potential efficacy of thalidomide during active disabling flare-ups, while evaluating duration, intensity, and frequency of flare-ups.” (Tenth Annual, 2001) While the study is ongoing, there is preliminary data available. By January 2001, there were 15 FOP patients involved in the study. The flare-ups continued with all patients taking thalidomide; however, patients and/or their parents reported that there was an improvement in the duration and severity of the flare-ups in 14 out of 15 of the patients. Since January 2001, seven patients had received their second annual bone scan. Of those seven, six showed no new site of heterotopic bone formation when compared to the initial bone scan at the beginning of the study. While the thalidomide trial continues and no definite results have been reported, this study has very promising preliminary data. (Treatment Considerations, 2001; Tenth Annual, 2001)

Retinoids are another area of interest for researchers searching for a treatment and cure for FOP. These agents are of interest because of their ability to differentiate connective tissue into cartilage and bone. A specific agent researchers have been looking into is Accutane, also known as isotretinoin or 13-cis retinoic acid. This medication is traditionally used in the treatment of severe acne. It is contraindicated in pregnant women because of the effects it has on the fetal skeleton. A clinical trial was conducted between 1984 and 1988 at the National Institutes of Health (NIH). Results indicated that growth of bone that had already began forming was not halted. It did appear to decrease new episodes of bone formation around joints by at least 27%. This study also indicated that isotretinoin had some unpleasant side effects. This agent is not a cure, but it can offer some beneficial results for those who decide to take it. (Treatment Considerations, 2001; Wagman, Kantanie, & Kaplan, 2001)
Conclusion

Fibrodysplasia Ossificans Progressiva is a rare devastating and debilitating disease. Even though it physically affects fewer than 200 known people worldwide, it also affects those family and friends who can only sit around as the person eventually becomes bound within his/her own skeleton. Therefore, a treatment and cure is needed. There are a small number of scientists and researchers dedicated to finding a cure for this genetic disorder. Hopefully one day we can learn how to prevent and treat these patients and free them from their skeletal prison.
Nursing Diagnosis: Impaired Physical Mobility related to decreased strength and endurance secondary to musculoskeletal impairment.

<table>
<thead>
<tr>
<th>Expected Outcomes</th>
<th>Interventions</th>
<th>Rationale</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Demonstrate use of adaptive equipment to increase mobility.</td>
<td>• Instruct patient how to use assistive devices (i.e. walker, bath bench, bedside commode, etc.) correctly.</td>
<td>• Passive range of motion exercises are contraindicated for patients with FOP.</td>
</tr>
<tr>
<td>• Use safety measures to minimize potential for injury.</td>
<td>• Educate patient about appropriate safety measures to decrease risk for injury.</td>
<td>• Physical therapy is contraindicated for patients with FOP.</td>
</tr>
<tr>
<td></td>
<td>• Encourage active range of motion exercises.</td>
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Rationale:
- Passive range of motion exercises are contraindicated for patients with FOP.
- Physical therapy is contraindicated for patients with FOP.
**Nursing Diagnosis:** Pain, acute, related to tissue trauma secondary to lesions associated with FOP.

<table>
<thead>
<tr>
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<tbody>
<tr>
<td>• Patient will state a decrease in pain.</td>
<td>• Administer prescribed medications, such as NSAIDs or Cox-2 inhibitors, for pain.</td>
<td>• Warm water of a whirlpool helps with ease the pain associated with flare-ups.</td>
</tr>
<tr>
<td></td>
<td>• Position patient for comfort.</td>
<td>• Using whirlpool on a gentle cycle helps to prevent injury/trauma to tissues leading to a second flare-up.</td>
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<td></td>
<td>• Acknowledge client’s complaint of pain as real.</td>
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<tr>
<td></td>
<td>• Encourage patient to use a whirlpool on a gentle cycle during flare-ups.</td>
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Nursing Diagnosis: Ineffective Breathing Pattern related to immobility secondary to decreased chest wall expansion.

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<tr>
<th>Expected Outcomes</th>
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<tr>
<td>• Patient will achieve maximum pulmonary function.</td>
<td>• Encourage deep breathing.</td>
<td>• Receiving an influenza vaccine will decrease complications of respiratory illness.</td>
</tr>
<tr>
<td>• Patient will remain free from respiratory illness.</td>
<td>• Encourage patient to receive annual influenza vaccine (subcutaneously) during the flu season.</td>
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<tr>
<td>• Patient will maintain respiratory rate within normal limits.</td>
<td>• Assess respiratory rate frequently.</td>
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</tr>
<tr>
<td>• Patient will maintain ABGs within normal limits.</td>
<td>• Monitor for abnormal ABGs and notify physician if values are abnormal.</td>
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**Nursing Diagnosis:** Risk for Injury related to altered mobility secondary to Fibrodysplasia Ossificans Progressiva.

<table>
<thead>
<tr>
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<th><strong>Rationale</strong></th>
</tr>
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<tbody>
<tr>
<td>• Patient will remain free from injury.</td>
<td>• Instruct patient to remove or decrease obstacles at home (i.e. throw rugs, clutter, etc.) that will increase the risk of injury.</td>
<td>• If a patient knows the correct way to use an assistive device, then he/she will be at a decreased risk of injuring himself/herself while using the device.</td>
</tr>
<tr>
<td>• Patient will implement safety measures to prevent falls and other injuries.</td>
<td>• Educate patient about correct way to use assistive devices.</td>
<td></td>
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<tr>
<td>• Patient will identify factors that increase risk for injury.</td>
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**Nursing Diagnosis:** Impaired Skin Integrity related to the effects of pressure and/or immobility secondary to motor deficits.

### Expected Outcomes
- Patient will identify causative factors for skin breakdown.
- Patient will notify care giver if change of position is needed.
- Patient will remain free from pressure ulcers.

### Interventions
- Assess skin frequently for signs and symptoms of skin breakdown.
- Reposition patient frequently (every two hours) and as needed.
- Keep moist areas (i.e. skin creases) dry.
- Place proper padding over any bony prominences.
- Implement wound care protocol or notify enterostomal nurse if pressure ulcer develops.

### Rationale
- Keeping moist areas dry will promote skin to stay intact.
- Repositioning prevents excessive pressure to one area.
**Nursing Diagnosis:** Altered Nutrition: Less than Body Requirements related to the inability to chew.

<table>
<thead>
<tr>
<th><strong>Expected Outcomes</strong></th>
<th><strong>Interventions</strong></th>
<th><strong>Rationale</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>• Patient will maintain body weight.</td>
<td>• Encourage patient to eat small frequent meals and snacks throughout the day.</td>
<td>• Eating small frequent meals and snacks throughout the day will help increase caloric intake.</td>
</tr>
<tr>
<td>• Patient will demonstrate no signs or symptoms of malnourishment or dehydration (i.e. dry skin, brittle nails, thin hair, dry mucous membranes, poor skin turgor, etc.).</td>
<td>• Teach patient about nutrient dense foods.</td>
<td>• Nutrient dense foods will decrease risk of malnutrition.</td>
</tr>
<tr>
<td>• Patient will verbalize food choices that are nutrient dense.</td>
<td>• Encourage patient to use dietary supplements (i.e. Ensure) as needed.</td>
<td>• Dietary supplements can help in meeting adequate caloric needs.</td>
</tr>
<tr>
<td>• Patient will identify foods that are easily eaten with a fused jaw.</td>
<td>• Discuss with patient foods that are easy to ingest or how to prepare foods that are easy to ingest with a fused jaw.</td>
<td>• Knowing what foods that are easy to eat will encourage adequate nutrition.</td>
</tr>
<tr>
<td></td>
<td>• Encourage adequate oral fluid intake.</td>
<td>• Adequate oral fluid intake will help prevent dehydration.</td>
</tr>
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<td></td>
<td>• Administer intravenous fluids as prescribed by physician.</td>
<td></td>
</tr>
</tbody>
</table>
Nursing Diagnosis: Body Image Disturbance related to changes in appearance secondary to chronic disease.

**Expected Outcomes**
- Patient will verbalize and demonstrate acceptance of appearance.

**Interventions**
- Encourage independence.
- Provide anticipatory guidance about the disease process of Fibrodysplasia Ossificans Progressiva.

**Rationale**
- Independence will give the patient the self-confidence needed to accept themselves.
- Providing anticipatory guidance about the disease process will give them the insight as what to expect and plan for in the upcoming years.
References


