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Rehabilitation in fibrodysplasia ossificans progressive - experience Report

Abstract

Introduction: Fibrodysplasia Ossificans Progressiva causes gradual limitation of joint range of motion by heterotopic osteogenesis in various connective tissue structures. It is a rare autosomal dominant disease and is correlated with the presence of congenital malformation of the bilateral hallux valgus, presence of ossification that appears spontaneously or precipitated by trauma.

Method: The search for the respective titles for FOP and Rehabilitation via a bibliographic review in the Pubmed database, the titles and abstracts were discriminated when they did not bring in their content the treatment of this disease.

Results and Discussion: The 9-year-old patient comes with complaints of pain in the joints of the shoulders, elbows, cervical and thoracic spine. Slow walking, structural deformities in the axial axis of the cervico-thoracic spine, decreased shoulder abduction up to 15°, limited elbow flexion and extension. The FOP treatment strategy is a comprehensive program of activities to prevent the myriad possibilities of trauma, from avoiding unnecessary surgical procedures, injections and even biopsies. Dry needling or acupuncture techniques are always contraindicated. Hydrotherapy turns out to be another useful tool in the process of preventing and relieving injuries, contributing to physical conditioning in a safe environment with low impact and cardiopulmonary performance. From the clinical observations, there will be indication of orthoses, auxiliary means for locomotion or adapted wheelchairs. Devices with voice command facilitate some daily activities, encouraging music therapy, dance or theater also favors the well-being of these patients.

Conclusion: The disease has some striking characteristics that favor early diagnosis. Injury prevention, medical management of acute painful flare-ups, and rehabilitation are pillars of treatment. There is still no curative treatment, however, the determination of a therapeutic plan prevents future trauma and can guarantee an adequate prognosis even though it is difficult and full of functional limitations for patients.

Keywords: fibrodysplasia ossificans progressiva, myositis ossificans, child, rehabilitation, cronic pain

Volume 8 Issue 1 - 2023

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Received: March 05, 2023 | **Published:** March 20, 2023

Abbreviations: FOP, fibrodysplasia ossificans progressiva; HPV, human papilloma virus; PCV, pneumococcal conjugate

Introduction

A disease as devastating as its former name of "stone man" syndrome, causes gradual joint limitation by progressive heterotopic osteogenesis in various connective tissue structures such as muscles, fascia, ligaments and tendons. Fibrodysplasia Ossificans Progressiva (FOP) is a rare autosomal dominant disease with evidence of the ACVR1/ALK2 gene mutation that activates aberrant formation of bone tissue and has a prevalence of less than 1 in a million inhabitants.

Osteoarticular mobility is increasingly impaired with functional disorganization of the range of motion of small and large joints, compromising the structure of the spine at an early stage, making walking difficult as the disease progresses and still lacks curative treatment, inevitably there is box restriction thoracic and pulmonary function favoring infectious diseases in addition to the high risk of falls.

There is usually a correlation with congenital malformation of bilateral hallux valgus and even early ossification, spontaneously or precipitated by trauma, such as intramuscular injections, symptoms such as pain and edema may precede the site where this bone formation will occur. It most typically occurs on the axial axis and posteriorly towards the ends. When it occurs on the face or neck, it can affect swallowing and speech. Although heterotopic ossification is by far the major feature of the disease, it can also course with impaired chondrogenesis and central nervous system dysregulation.

In this discussion, we will bring the considerations of the challenging rehabilitation, within the physiatric point of view, for a nine-year-old patient, with five years of FOP evolution, obtaining limitations already established in the cervical spine, shoulders, elbows and fingers, which considerably limit the various activities and functionalities of the patient during her day.

Material and methods

The respective titles of MeSH Terms were searched within the Pubmed database, defined as ("Habilitation" OR "Sports for Persons with Disabilities" OR "Recovery of Function" OR "Rehabilitation" OR "Exercise" OR "Physical Therapy Modalities") AND ("Myositis Ossificans" OR "Fibrodysplasia Ossificans Progressive" OR "Progressive Myositis Ossificans" OR "Progressive Ossifying Myositis" OR "Myositis Ossificans Progressive"). A total of 180 articles were found and discriminated according to exclusion criteria based on titles and reading of abstracts or did not include part of the treatment of this disease in their content. Our case report served as the basis for a review of the rehabilitation literature on FOP rehabilitation.

Results

A 9-year-old Caucasian female patient, being followed up by the Physiatrics team at the Institute of Physical Medicine and Rehabilitation, comes to the outpatient clinic with complaints of pain in the shoulder joints, elbows, cervical and thoracic spine. Presenting limitation for trunk flexion and extension, with limitation of range of motion in shoulder elevation. Apparently healthy until the age of 4, when the investigation of the disease began in another hospital with subsequent follow-up at Hospital das Clínicas de São Paulo, with several specialties, since she had persistent joint pain in the extension of the neck with irradiation to the head and shoulders, wrist pain, limited circumvolution of the shoulder, signs of axial rigidity and gait deficit.

Before the FOB diagnosis, the 2-year-old patient underwent orthopedic surgery on the bilateral hallux. She uses daily medications such as low-dose prednisone, baclofen, gabapentin, lactulose, omeprazole, dipyrone and naproxen during pain crises. It claims points of pain relief when lying down. Associated with the motor limitation, the patient also had a delay in cognitive development for her age, with difficulty keeping up with tasks with the class at school. He uses public transport frequently, including to go to school, there he cannot extend his neck to see the lessons on the blackboard among other activities. He is currently carrying out therapies with a respiratory and motor physiotherapy team, in addition to activities with an Occupational Therapy, Psychology, Speech Therapy and Nursing team.

Still no indication for the use of orthoses, independent walking with slow gait with forefoot support both at home and in the community. Occasionally has falls, with the last 6 months before the consultation. Limitation for postural changes and transfers, as he has been presenting worsening of trunk control to get up or sit down from chairs, needing help not to lose his balance. Needs help in bathing to wash the head, for intimate hygiene, for dressing the upper and lower limbs. Can eat alone when not in pain crises.

On physical examination, the patient was in good general condition, static neck position with limitation of all movements associated with disproportionate anterior position and slight downward tilt. Deformities felt in the axial axis of the cervico-thoracic spine, decreased shoulder abduction of up to 15°, flexion and extension of limited curvature, but reducible. Showing shortening of the sural triceps with restricted dorsi and plantar flexion of the feet with the presence of bilateral hallux valgus. Showing grade five strength overall except for grade 4 in bilateral dorsiflexion.

Discussion

From the suspicion of the diagnosis, with the hallux valgus malformation, short thumbs, hypoplasia of the digital phalanges, fusion of cervical vertebrae, heterotopic ossifications and soon when confirmed by the identification of a variant of the ACVR1/ALK2 gene, the way of treating FOP becomes a broad program of preventive activities for countless possibilities of trauma, from avoiding unnecessary surgical procedures, injections or biopsies.

In some situations, prophylactic corticotherapy is used, since the inflammatory cascade can contribute to musculoskeletal pain and edema. During flare-ups and crises of painful inflammation, in addition to corticosteroids, non-steroidal anti-inflammatory drugs and bisphosphonates are needed in short cycles. Several promising medications for the treatment involve gene therapy as a mechanism to inhibit the ossification protein activation cascade via the Activin A receptor.

A fall prevention trend and the use of helmets represent essential interventions for care management. Relief options with physical means techniques such as warm water can be useful for pain control. Dry needling or acupuncture techniques are always contraindicated. Hydrotherapy turns out to be another useful tool in the process of preventing and relieving injuries, contributing to physical conditioning in a safe low-impact environment and cardiopulmonary performance. Iontophoresis involving the introduction of physiologically active ions applied topically may help restore some of the range of motion lost to the temporomandibular joint in FOP.

Fortunately for most children with FOP, they will have a complete immunization schedule prior to diagnosis. Although immunization by subcutaneous administration is recommended for all vaccines that can be administered this way. For other vaccines, the risk/benefit of intramuscular administration should be discussed with parents. Considering that immunizations should not be carried out during outbreaks. Some vaccines that can only be administered intramuscularly because SC or intradermal application can cause severe local irritation, induration, skin discoloration, inflammation and granuloma formation, are as follows: Diphtheria, tetanus, pertussis (DTaP, DT, Tdap, Td); Haemophilus influenzae type b (Hib); Hepatitis A (HepA); Hepatitis B (HepB); Human Papilloma Virus (HPV) and Pneumococcal Conjugate (PCV). An intranasal flu vaccine is available for administration when there are no contraindications in individuals 5 to 49 years of age.

Early treatment will help slow the progression of the disease and provide children with a better quality of life. As the condition progresses, the disease leads to joint ankylosis, and the aggravating factors promote breathing difficulties and restricted mouth opening, and can shorten the patient's lifespan. Even with some differential diagnoses of other musculoskeletal diseases, it was not evident that the ACVR1 mutation does not have an oncogenic role. Recurrent severe headaches associated with tension-type or migraine headaches are not uncommon, in addition to neurological manifestations such as neuropathic pain, allodynia and hyperalgesia.

Patients in the adult phase usually lose functionality and mobility becomes restricted, limited to bed or chair around the age of 30. Due to the chance of a fused spine, fixed pelvis, with limited hip and knee range of motion as well as foot dorsi and plantar flexion. As heterotopic bone formation is permanent and leads to progressive immobility, the patient's tendency is to increase assistance in carrying out activities of daily living. Patients who previously had modified independence now have semi-dependence or complete dependence for BADLs and IADLs. The disease progresses restrictively, affecting the chest wall, and secondary heart failure leads to death, commonly in the fifth decade.

Dental procedures are sometimes avoidable in order not to favor limitation of the pterygoid musculature, temporomandibular ankylosis and mandibular hypoplasia, as they may favor obstructive sleep apnea and pulmonary hypertension. Prevention of tooth decay is essential to avoid the need for more invasive treatment. In the literature, we also see confirmation of the effects of post-COVID-19 causing uncontrolled seizures in a patient with FOP.

Functionality questionnaires such as the Barthel Index indicate a need for parameterization and follow-up related to the first reductions in ABVDS. With the objectives of educating about best practices for care, sharing valuable clinical experiences from the care of patients with classic FOP and variants, a training committee of experts in the area proposed to discuss the main advances for the scientific community.

Citation: da Costa TR, Helfenstein T, Amaral AAO. Rehabilitation in fibrodysplasia ossificans progressive - experience Report. *Int Phys Med Rehab J.* 2023;8(1):95–98. DOI: 10.15406/ipmrj.2023.08.00340

Activities with a high risk of injury or falls such as running, cycling or contact sports should also be avoided as far as possible, an individualized lifestyle plan should be tailored based on age, limited mobility and cultural norms. The patient's and family's knowledge, values, beliefs and cultural origins must be incorporated into the planning and delivery of patient care, valuing the patient-centered view. The physician must communicate and share complete and unbiased information with patients and families in an affirmative and useful manner and to allow the patient to participate effectively in the decision-making process. As with any rare disease, a multidisciplinary approach to comprehensive medical care and social support from different specialists is advisable.

FOP is often misdiagnosed as fibromatosis, desmoid tumor or cancer, bunion, myositis, arthritis and rheumatic diseases. Delay in diagnosis not only delays management, but may also expose the patient to unnecessary interventions. In addition, early diagnosis can also make the patient aware of the precautions to be taken. The care taken to maintain a management approach that is not guided by a specialized team can compromise the ROM of the joints and even vigorous physical exercise impacts on the pathophysiology and further limits the patient.

Rehabilitation challenges range from promoting independence to preserving energy in the face of daily limitations. Occupational Therapy, Physiotherapy and Speech Therapy teams are fundamental in the follow-up of these patients. From adaptations for food with change of consistencies or even utensils for practicality and greater autonomy can be used, and even training for appropriate clothing or footwear given the limitation of ROM of limbs and spine. Adaptations in the rooms, especially in the bathroom, may be necessary, in addition to the adequate height of seats and tables, with less functional impairment.

Based on clinical observations, the use of orthoses, auxiliary means for locomotion or even the need for wheelchairs with adaptations for cervical support, activity table or reclining backrest. Adaptations both at home and at school favor social insertion, broaden the horizon of access and instruction of the patient who will sometimes need special transport. The practicality of current technology and devices with remote or voice commands reduce the distance from some daily activities.

For adult patients, sexual practice also requires care, as it allows unusual positions for non-flexible joints. Including guidance regarding reproduction in the case of women, contraception and genetic counseling, it is a duty of the care team to guarantee the relevant guidance.

Recreational activities are encouraged by professional psychologists and social workers. This support for patients and families minimizes the inconveniences imposed by FOP. Other therapies such as art therapy, music therapy, dance and theater are tools in creative processes with the aim of improving deficiencies by favoring well-being.[1-15]

Conclusion

Even though it is rare, the disease has some striking characteristics that favor early diagnosis. The help of genetics is crucial to characterize FOP and assertively determine adequate prevention, therapy and rehabilitation. Diagnoses can be neglected or delayed, resulting in possible biopsies and other unnecessary interventions, which favor the worsening of the disease course. Injury prevention, medical management of acute painful flare-ups and rehabilitation are the 3 pillars of treatment.

Training, guidance and alerting physicians, multi-team professionals and especially family members are essential for improving the care of these patients, guaranteeing quality of life within the possibilities of motor rehabilitation. Even if there is no curative treatment, the determination of a therapeutic plan prevents future traumas and guarantees part of the adequate prognosis, even if difficult and full of functional limitations. Our case illustrates an early physiatric intervention where a creative rehabilitation allows to unleash the human potential of people with FOP.

Acknowledgments

This article could not have been done without the support of the multidisciplinary team that assists the entire physiatric team, thank you all.

Conflicts of interest

Declare there are no conflicts of interest.

Funding

None.

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Citation: da Costa TR, Helfenstein T, Amaral AAO. Rehabilitation in fibrodysplasia ossificans progressive - experience Report. *Int Phys Med Rehab J.* 2023;8(1):95–98. DOI: 10.15406/ipmrj.2023.08.00340