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Review Article

Paediatric Hypermobile Connective Disorders and Juvenile Idiopathic Arthritis: Early Diagnosis, Occupational Therapy and Physiotherapy Perspectives

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Received: April 26, 2022; **Published:** June 28, 2022

Abstract

There is a greater need for an increased awareness of Paediatric rheumatic and musculoskeletal disorders (RMD) since failure to recognise symptoms and diagnose appropriately RMDs can result in long term disabilities and reduced mortality in children with inflammatory RMDs. Over the years, it has become apparent that early diagnosis and targeted management plans result in very out disease outcomes. No-pharmacologic treatments are also very important and should be implemented soon after diagnosis. We review the current occupational therapy and physiotherapy management modalities in children with inflammatory and non-inflammatory RMDs.

Keywords: *Hypermobile EDS; EDS; JIA; Juvenile Idiopathic Arthritis; Inflammatory RMD; Paediatric Occupational Therapy*

Introduction

Joint pain is a common rheumatic and musculoskeletal symptom in children which may be acute or chronic and may be related to joint inflammation. Missing joint inflammation may result in irreversible long term deleterious consequences in children. The commonest chronic inflammatory rheumatic and musculoskeletal disease (disorder) {iRMD} is termed juvenile idiopathic arthritis (JIA). Its diagnosis in children may be elusive. However, noninflammatory RMDs including hypermobile connective disorders are a more common cause of joint pain. The purpose of this review is to focus and highlight the iRMDs and non-inflammatory hypermobile connective tissue joint disorders, the importance of early correct diagnosis, early intervention and the role played by physiotherapy and occupational therapy in the management of these disorders in addition to the specific targeted management of iRMDs.

JIA

The ILAR classification system defines JIA as all forms of inflammation of one or more joints beginning in children younger than age 16 years and inflammation in at least one joint persisting for longer than 6 weeks. Other causes of joint inflammation such as infection should

be excluded [3]. JIA is further classified into seven categories based on inclusion and exclusion criteria according to features present within the first six months of disease (Table 1). The disease subtype should be assessed at the onset of the disease and reviewed subsequently at each clinic visit. The development of new clinical features during the course of the disease determines the final disease subtype.

1. Polyarthritis RF positive
2. Polyarthritis RF negative
3. Oligoarthritis
a. Persistent
b. Extended
4. Psoriatic arthritis (JPsA)
5. Enthesitis-related arthritis
6. Undifferentiated arthritis
7. systemic-onset JIA (sJIA)

Table 1

JIA can develop at any age but very rarely in infants up to 6 months of age. The incidence in girls is twice that in boys suggesting a role of hormonal factors. The triggers of JIA are unknown. Immunogenetic, environmental, infectious and epigenetic factors are most likely involved. The pathophysiologic mechanisms in JIA include inflammatory cytokines such as TNFα, IL-1 β and IL-6 as well as macrophages and T cells. These inflammatory pathways have are provided therapeutic targets that target specific cytokine and cell pathways. By contrast, Systemic juvenile idiopathic arthritis (sJIA) has a different clinical profile characterised systemic features with spiking fevers, skin rash, serositis lipid dysregulation and hepatitis as well as macrophage activation syndrome (MAS). The cytokine and cellular profile comprising of high levels of IL-1β, IL-6 and IL-18, and neutrophils and monocytes/macrophages rather than lymphocytes) suggest that the innate immune system is involved in its pathophysiology. Hence, sJIA is an auto-inflammatory disorder rather than a 'classic' autoimmune disease. The cytokine profile targeting has led to the use of the biodrugs Anakinra and tocilizumab that inhibit IL-1β, IL-6 respectively. These interventions are highly successful in controlling its disease activity.

Undiagnosed and untreated, chronic inflammation in joints leads to cartilage destruction, impaired epiphyseal end plates growth, with marked reduction in joint function and activities of daily living [1]. Many other extra-articular complications may arise including growth retardation, uveitis, blindness, amyloidosis and life-threatening macrophage activation syndrome (MAS) [1]. Some of the complications are drug-side effect related e.g. osteoporosis, growth retardation secondary to glucocorticoids and chronic disease [2]. It is therefore imperative that early correct diagnosis is established and early appropriate aggressive treatment initiated. The aim of the therapy should be multidimensional: to control inflammation and pain, to preserve the range of motion, muscle mass and muscle strength, to induce disease remission, manage systemic complications and facilitate normal physical and psychosocial development. The drug options comprise of non-biologic disease modifying drugs {DMARDs} such as Methotrexate a Folate antagonist; Sulphasalazine which inhibits dihydropteroate synthase; Leflunomide which inhibits dihydroorotate dehydrogenase and Cyclosporine-A that works by inhibiting calcineurin. A number of biologic DMARDs {TNF α suppressors - Etanercept, Infliximab, Adalimumab; IL-1β targeted biodrugs - Anakinra, Canakinumab, Rilonacept; anti-IL-6 drugs - Tocilizumab and Sarilumab; T cell function inhibition - Abatacept; and anti-CD20 targeted Rituximab}. This mini review will focus on the non-drug interventions of occupational therapy and physiotherapy.

Hypermobility in children

Hypermobile connective tissues disorders may cause joint pain in children. New classification criteria were developed in 2017 to differentiate benign joint hypermobility now called hypermobile EDS (hEDS) where the Beighton score is over 5/9 and if less Hypermobile

spectrum disorder (HSD), from the more serious genetic associated hypermobile connective tissue disorders such as Ehlers-Danlos syndrome (EDS) subtypes, osteogenesis imperfecta, Marfan syndrome and homocysteinaemia among others. The clinical features of hEDS are similar to some extent to these disorders. Genetic testing may be necessary to establish the exact diagnosis. It is important to distinguish the non-inflammatory causes of joint pain in children from the inflammatory joint disorders. In the early years of development, most children are hypermobile and therefore a diagnosis of hypermobility as a cause of joint pain should not be made before the age of 5 years at least. Referral to a Paediatric Rheumatologist with expertise in hypermobile connective tissue disorders is vital. Early correct diagnosis and intervention are again key to good outcomes in children with hypermobility and arthralgia.

Occupational therapy

Physiotherapy and occupational therapy are valuable components in the comprehensive management of RMDs in children to maintain core stability, physical fitness and overall motor functionality. It is however very important to distinguish inflammatory arthritis {active and non-active} and Hypermobile connective disorders so that an appropriate management plan is initiated.

Occupational therapy provides a wide range of interactions targeted at improving and enhancing motor skills, self-reliance and independence; improving physical and cognitive functions and fostering self-reliance in order to ensure optimal quality of life. OT also helps children with RMDs to participate in school activities. The over-arching other objectives of OT are to restore and maintain total function in areas such as activities of daily living, work, and leisure in people who have lost the ability to care for themselves, work or perform their daily duties and life roles. OT will assist children to adapt to everyday life so that they can achieve independence and autonomy in their functions.

The importance of OT was demonstrated in a study Pozmohova., *et al*. (2021) in Ukraine who confirmed that early occupational therapy intervention enhances the likelihood of maintaining function in rheumatoid arthritis patients [4]. After three months of occupational therapy, there was a marked improvement in the compression force of the wrist in comparison to the patients who were in the control group [4]. Similar observations have been made in JIA [9].

During the inflammatory phase of inflammatory arthritis, the main objectives of rehabilitation for patients with iRMD are to reduce pain, preserve the functionality of the affected joints, the ability to self- care, and perform household duties, professional work, and stabilization of the affected joints. The aim is also to improve quality of life of the patient. Early occupational therapy is recommended to enhance and maintain function and mobility.

During the non- inflammatory phase, intervention includes increasing muscle strength, reducing pain and increasing tolerance to exercise. Occupational therapy is essential in the treatment of JIA as occupational therapists train in motor function, motor skills, activities of daily living, performing exercises with less pain, joint protection, use of helpers and splints [4].

Using a splint improves the force of compression of the wrists, reduced joint pain, but limits range of motion. Training patients on joint protection enables patients to tolerate activities of daily living such as household chores, dressing, and cooking with reduced pain [4].

Seigel., *et al*. performed a literature review on 51 studies and found strong evidence that supports the use of aerobic exercises, resistive exercises and aquatic therapy [5]. Among the psycho-education interventions, strong evidence supports the use of patient education, self-management, cognitive - behavioural approaches, multi- disciplinary approaches and joint protection [5]. There was limited evidence for the use of assistive technology and emotional disclosure [5]. The evidence supports the use of occupational therapy interventions, but few interventions were occupation based [5].

Literature on Occupational therapy interventions in children with JIA in Africa is unavailable. There is therefore a need for more research to be performed regarding the management of inflammatory arthritis in Africa. There also needs to be more awareness of conditions of inflammatory arthritis in Africa.

Occupational therapy perspectives in hypermobile EDS and HSD

In hypermobile connective tissue disorders, Occupational therapy and physiotherapy interventions are often targeted at self- management, enhancing muscle strength and sense of proprioception in the affected joints and limbs [6]. We review the occupational therapy and physiotherapy perspectives on the management of hEDS and HSD.

Hakim., *et al*. (2017) recommend a multi- disciplinary approach to pain and fatigue in managing hypermobility [6]. Early diagnosis of hEDS is important to address chronic pain, fatigue and occupational performance before developing a disability [7]. Occupational therapists teach adapting strategies to enhance function [8]. These adaptations include pacing, changing sleep patterns, support to change jobs or hours to work, adaptation of the environment and assistive devices and gaining more independence [6,9].

Occupational therapists have a holistic approach to find ways for self- management and exercises [10]. Occupational therapists teach relaxation techniques to help manage stress and pain, graded exercise therapy, client centred goal setting, training coping skills to manage setbacks; planning and prioritizing activities of daily living and splitting activities into smaller, manageable tasks [10]. Occupational therapists perform school visits and the assess any challenges the child with hypermobility may have in their environment such as functional mobility, hand writing and performance in physical education and school attendance [9].

The important role that occupational therapists play with patients with hypermobility are pain and fatigue education, organizing living according to available energy, supporting occupational choices and assisting in balancing between activity and rest [11]. A study conducted by Nahi (2020) in Finland found that pain and fatigue affected the lives of patients with Ehlers- Danlos Hypermobility type and they experience severe fatigue and episodes of severe pain which adversely affect performance in activities of daily living, routines, occupational choices as well as the social and physical environment [11]. Most participants reported cycles of musculoskeletal injuries and problems with their joints that affected that affected their physical capacity in various ways. All participants suffered from joint pain in their fingers and in their wrists and had challenges in adapting to physical hand - related activities such as taking down groceries from the shelf [14]. The participants also had their own means to manage their pain and fatigue such as putting their hands in cold water, shaking and massaging them [11]. Hypermobility led to the reduction of participation in activities that required a lot of expenditure of energy such as ball games, gymnastics or running [11]. For finger and wrist pain, Occupational therapists worked on joint control [11]. For impaired body coordination occupational therapists manufactured orthosis and pressure garments. If patients had challenges in fine motor activities, the occupational therapists advised that they should pause the activity. For activities requiring long- term standing and sitting, muscle training was performed [11]. The occupational therapists engaged the patients in low energy exercises to enhance the patients' ability to participate in physical activities. Adaptation of the physical environment through use of proper ergonomic principles and use of assistive devices was used to adjust to a physically challenging environment and for energy conservation. Adapting working hours and work patterns and retaining of leisure activities was performed for energy conservation (1). Occupational therapists can manage pain and fatigue by stabilizing loose joints with bracing and taping, improving ergonomics at home and in the workplace [11]. OT can ameliorate pain and fatigue education; learning to organize living according to available energy, supporting occupational choices and assisting in balancing between activity and rest [12]. However, Nahi (2020) commented that fatigue is not being adequately addressed in patients with hypermobility [12]. Literature on the occupational therapy and physiotherapy interventions of hypermobility in Zimbabwe and in Africa in general are scarce and more research and awareness of conditions of hypermobility in the African context is important in order to formulate contextually relevant management strategies.

Physiotherapy perspectives

It is important again to distinguish inflammatory arthritis versus Hypermobile connective disorders. Physiotherapy should be initiated from the time the exact medical diagnosis is made, regardless of the active or non-active status of the disease. Overall, the main objectives of physiotherapy are to prevent long term consequences and sequelae of RMDs by reducing loss of musculoskeletal function, elimination of joint or back pain, maintaining the range of movement, and correction of any existing joint deformity. A variety of techniques that are passive including hydrotherapy and active are employed after assessing the child and planning the management pathway.

In Inflammatory arthritis, the aim of physiotherapy is to reduce pain and stiffness in the affected joint, restore correct posture and enhance quality of life [13]. Physiotherapists engage patients with inflammatory arthritis in range of motion (ROM) exercises, muscle strengthening and aerobic exercises [13]. ROM exercises are useful for maintaining joint flexibility and good posture [13]. Training proper positioning during sleep and properly aligned posture when walking [13]. Back and abdominal exercises also assist in maintaining an upright posture [13]. Physiotherapists also engage patients in regular physical activity such as Tai Chii, Pilates and swimming, which can improve the overall health and wellbeing of the patient of the patient [13]. Exercise therapy is advantageous as it offers an aerobic workout, promotes spine flexibility, physical endurance and reduces the risk of cardiopulmonary complications [13]. Another modality, Ultrasound therapy and aquatic therapy have also been shown to be effective in relieving pain and enhancing function [15].

Physiotherapy is also very essential in children diagnosed with HSD/ hEDS. If physiotherapy is implemented correctly, it assists in pain reduction, and improving endurance, functional prognosis as well as quality of lives of people with HSD/hEDS [16]. Physiotherapy interventions include patient and caregiver intervention, exercises the patient can tolerate, pain, fatigue and stress management, self- management techniques and splinting or bracing recommendations, cognitive behavioural strategies and referral to other health professionals where necessary [16]. Patients may also benefit from neuro- muscular re- education, therapeutic exercises, manual therapy, and patient education about injury prevention, body mechanics and joint prevention [16]. Compression clothing, tapping and orthotics may improve proprioception in patients [16]. Proprioceptive training may enhance movement precision and fear of movement found in this population [16]. Muscle strengthening is important in reducing muscle pain. Muscle strengthening exercises should be gradual. Joint stability can be addressed through postural awareness, proper body mechanics, muscle strengthening and motor control training and consistent physical activity to improve physical fitness [16]. Spinal stabilization exercises can reduce pain and enhance overall function. Weight management is important, especially for younger patients [16].

Finally, fatigue is a common feature in all forms of RMDs [17]. Fatigues can be addressed through patient education and exercise. All patients must take at least Vit D3 2000Units per day. Patients should be educated on sleep hygiene, relaxation and stress management techniques, pacing and prioritising. Muscle strengthening can also reduce fatigue [17]. Evidence for the effectiveness of physiotherapy in the treatment of HSD/ hEDS has not been systematically studied and more work is needed to establish its exact role [16].

Conclusion

Making an early exact diagnosis and initiating an appropriate management plan both pharmacological, surgical and non-pharmacologic interventions are critical to ensuring good clinical outcomes in children with JIA and hypermobile connective tissue disorders. Targeted biodrug approaches have proved highly successful in the management of childhood iRMDs. A wide range of OT and physiotherapy management modalities are available that give children with RMDs a chance to maintain and achieve self-reliance and independence in the future.

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Volume 11 Issue 7 July 2022