Narrative Review

Beyond Bleeds: Restoring Function in Haemophilia

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Abstract

Haemophilia is an inherited bleeding disorder with significant musculoskeletal and functional consequences. Although factor replacement therapy remains the cornerstone of management, the role of rehabilitation in preventing disability and improving long-term functional outcomes is increasingly recognised. Recurrent hemarthroses can lead to chronic synovitis, haemophilic arthropathy, pain, muscle weakness, gait abnormalities, and reduced participation in daily activities. This narrative review summarises the pathophysiology of musculoskeletal involvement in haemophilia, common functional impairments, rehabilitation assessment, exercise therapy, physical modalities, orthotic management, and multidisciplinary rehabilitation strategies. The review aims to provide a concise and clinically relevant overview for Physiatrists and rehabilitation professionals involved in haemophilia care.

Introduction

Haemophilia is a rare inherited X-linked recessive bleeding disorder caused by deficiency of clotting factors. Haemophilia A results from factor VIII deficiency, whereas the less common haemophilia B is caused by factor IX deficiency. Individuals with haemophilia experience prolonged bleeding because of inadequate clotting factor activity.

Traditionally, haemophilia was considered a disorder affecting only males, while female carriers were believed to remain asymptomatic. However, it is now recognised that many women and girls carrying the haemophilia gene may also experience bleeding manifestations[1].

It is estimated that approximately 21 per 100,000 males have haemophilia A and around 4 per 100,000 males have haemophilia B. In Kerala, this corresponds to nearly 3600 persons with haemophilia (PWH)[2]. Based on factor activity levels, haemophilia is classified as:

- Mild: 5% - 40% of normal factor activity
- Moderate: 1- 5% normal factor activity
- Severe: < 1% normal factor activity

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Clinical manifestations include:

- Easy bruising
- Prolonged bleeding after injury
- Bleeding into muscles and joints
- Spontaneous bleeding episodes

Bleeding into joints or muscles may produce:

- Pain or discomfort
- Swelling and warmth
- Joint stiffness
- Difficulty using the affected limb

Musculoskeletal bleeding is the hallmark manifestation of haemophilia and contributes substantially to morbidity and long-term disability. Recurrent hemarthroses involving the knees, ankles, and elbows may lead to synovial hypertrophy, cartilage destruction, deformities, chronic pain, muscle wasting, impaired mobility, and reduced quality of life.

Although haemophilia management primarily focuses on factor replacement therapy, increasing recognition of chronic musculoskeletal impairment has highlighted the importance of rehabilitation in comprehensive haemophilia care. Rehabilitation aims not only to restore function after bleeding episodes but also to prevent disability, optimise mobility, improve participation, and enhance psychosocial well-being.

This narrative review discusses the role of rehabilitation in haemophilia, with emphasis on functional restoration, prevention of disability, and rehabilitation strategies across different stages of the disease.

Pathophysiology of Musculoskeletal Involvement

The development of haemophilic arthropathy typically progresses through three stages:[3]

1. Acute hemarthrosis
2. Chronic synovitis
3. Degenerative arthritis

The synovium has limited ability to clear blood following repeated joint bleeds. Accumulation of blood breakdown products, particularly iron-containing hemosiderin, promotes synovial inflammation and proliferation. Pro-inflammatory cytokines including interleukin-1, interleukin-6, and tumour necrosis factor-alpha contribute to chronic synovitis and joint destruction[4–6].

The synovium gradually becomes hypertrophic and highly vascular, increasing susceptibility to recurrent bleeding and creating a vicious cycle of hemarthrosis and inflammation. Inflammatory mediators also interfere with cartilage maintenance, resulting in progressive cartilage degeneration[4,5].

The pathological changes in haemophilic arthropathy share similarities with both rheumatoid arthritis and osteoarthritis. Persistent inflammation, cartilage degeneration, deformity, and joint destruction eventually lead to chronic disability and impaired function.

Musculoskeletal Complications of Haemophilia

Musculoskeletal complications include:

- Recurrent hemarthrosis
- Chronic synovitis
- Flexion deformities
- Growth abnormalities
- Cartilage damage
- Haemophilic arthropathy
- Muscle wasting
- Gait abnormalities
- Functional impairment

The joints most commonly affected are the ankle, knee, elbow, and hip [7–9].

Pain and recurrent bleeding may lead initially to correctable deformities, which can later become fixed. Muscle weakness, altered biomechanics, and reduced physical activity further contribute to functional decline. Fear of bleeding often results in reduced participation in physical activity, leading to deconditioning, obesity, osteoporosis, and social isolation.

Functional Impairments In Haemophilia

Functional impairment in haemophilia is multifactorial and depends on bleeding severity, frequency of hemarthroses, treatment access, and rehabilitation availability. Reduced mobility may result from chronic pain, synovitis, arthropathy, restricted range of motion, muscle weakness and secondary musculoskeletal strain

Patients may develop antalgic gait, reduced stride length, toe walking due to ankle contractures, and compensatory movement patterns. Children may avoid sports participation, while adults may experience occupational limitations and psychosocial distress.

Rehabilitation Assessment

A comprehensive rehabilitation assessment should include evaluation of the target joints, muscle strength, range of motion, posture, balance, gait patterns, and functional abilities. The Hemophilia Joint Health Score (HJHS) is commonly used to assess joint involvement, particularly in the elbows, knees, and ankles[10]. The Functional Independence Score in Hemophilia (FISH) is a performance-based tool that evaluates the impact of musculoskeletal impairment on daily functioning and assists in planning rehabilitation interventions[11]. Imaging modalities such as ultrasound and MRI may help detect synovitis and early joint damage.

Role of Physical Medicine And Rehabilitation

Physical Medicine and Rehabilitation plays an important role in the prevention and management of disability in haemophilia. Rehabilitation aims to maintain functional independence, reduce participation restrictions, and improve quality of life.

Optimal haemophilia care requires a multidisciplinary team consisting of Haematologists, Physiatrists, Physiotherapists, Occupational Therapists, Orthopaedic Surgeons, Psychologists, Social Workers and Nurses. Collaborative care improves treatment adherence, reduces complications, and enhances functional outcomes.

Under appropriate haemostatic control, individuals with haemophilia can achieve good physical conditioning and participate safely in physical activity and daily life[12].

Rehabilitation goals include:

- Preventing recurrent bleeds and deformities
- Preserving joint integrity
- Restoring mobility and strength
- Improving balance and proprioception
- Managing pain
- Enhancing participation in daily activities
- Improving quality of life

Rehabilitation should be individualised according to age, disease severity, bleeding status, and functional goals.

Exercise Therapy

Exercise therapy is a core component of rehabilitation in haemophilia. Earlier concerns regarding exercise-induced bleeding have gradually shifted toward supervised and individualised exercise prescription.

Exercise offers several important benefits, including improved muscle strength, better cardiovascular fitness, enhanced joint stability, improved proprioception and balance, effective weight management, and improved psychological well-being.

Types of exercises commonly recommended include:

- **Range of Motion Exercises :** Gentle exercises help maintain flexibility and prevent contractures.
- **Strengthening Exercises :** Progressive resistance training improves periarticular muscle support and joint stability.
- **Aerobic Training :** Low-impact activities such as walking, cycling, and swimming improve endurance and cardiovascular health.
- **Proprioceptive Training :** Balance training and neuromuscular exercises improve joint awareness and may reduce injury risk.
- **Flexibility Exercises :** Stretching exercises help reduce stiffness and improve functional mobility.

Important considerations before prescribing exercise include bleeding risk, joint status, timing of factor replacement, presence of inhibitors, and the patient's pain levels. Exercise programmes should begin gradually and progress according to patient tolerance and clinical status[1].

Care should be taken to avoid overexertion, especially in individuals with active joint disease or recent bleeding episodes. High-impact and contact sports should be approached cautiously[13]. Proprioceptive rehabilitation is particularly important, as impaired joint proprioception may increase the risk of recurrent injury and instability.

A professionally supervised and individualised exercise programme has been shown to be feasible, safe, and beneficial in people with haemophilia, including those with haemophilic arthropathy[14].

Physical Modalities and Interventional Procedures

Physical modalities such as Transcutaneous Electrical Nerve Stimulation (TENS) may be used for pain management in selected patients. Orthoses and shoe insoles may help reduce pain and improve biomechanical alignment in individuals with arthropathy[15]. However, rigid orthotic devices should be used cautiously in unstable joints[16]. Arthrocentesis may occasionally be performed in acute, tense knee under adequate haemostatic cover to reduce pain and intra-articular pressure[15]. Patients with chronic synovitis and recurrent bleeding refractory to medical management may benefit from synoviorthesis or synovectomy. Radiosynovectomy has shown promising outcomes in selected cases[17].

Conclusion

Haemophilia is no longer viewed solely as a bleeding disorder but as a chronic condition with important musculoskeletal and functional consequences. Advances in medical management have improved survival and shifted attention toward long-term functional independence and quality of life.

Rehabilitation plays a central role in preventing disability, optimising mobility, reducing pain, and improving participation across the lifespan. Early intervention, patient education, structured exercise programmes, and coordinated multidisciplinary care can substantially improve outcomes in individuals with haemophilia. A comprehensive and individualised rehabilitation approach is essential to help patients move beyond bleeds towards meaningful functional recovery.

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