



IJCRR

Section: Healthcare

ISI Impact Factor  
(2019-20): 1.628

IC Value (2019): 90.81

SJIF (2020) = 7.893



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# The Efficacy of Orthotic Management in Haemophilia: A Narrative Review of Clinical Implication and Scope of Improvement

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## ABSTRACT

The patients with haemophilia disorder of blood clotting system characterized by excessive bleeding at the site of trauma and surgery and bleeding into soft tissue and joints, which may result in disability due to changing bony structure and cartilage destruction. Both haemarthrosis and hemophilic arthropathy (HA) badly affect the gait pattern and ultimately quality of life of a hemophilic patient. The objective is to conduct the narrative review and synthesis of published literature to highlight the current trends of treatment in hemophilic haemarthrosis and arthropathy and narrate the need of future trends and demand to uplift the user's experience and quality of life. A cross-sectional randomized literature search in PubMed, Wiley, Research Gate, Google Scholar, etc done and articles synthesized to extract the line of orthotic treatment in haemophilia and its efficacy. Although there is limited literature about the orthotic treatment in hemophilic patients are available, the results of various orthosis implication in hemarthrosis and arthropathy are encouraging. Despite the complication like synovitis and arthropathy in haemophilia are manageable today, there is a need to look into the changes in orthotic practice consideration, policy development, and further research development targeting the inclusive well-being of a hemophilic patient.

**Key Words:** Haemophilia A, Blood coagulation disorders, Orthotic devices, Foot orthoses, Rehabilitation, Joint diseases

## INTRODUCTION

Haemophilia is an X-linked genetic disorder of the blood clotting system characterized by excessive bleeding at the site of trauma and surgery and bleeding into soft tissue and joints, causing synovial inflammation, iron deposition, cartilage destruction, and bony changes. These arthritic changes cause pain and alter the activity of daily living-altering the movement. In individuals with arthropathy, reported changes from controls to include increases in swing time, stance time, double support, single support, and base of support. Decreases from controls include step length and normalized velocity.<sup>1</sup> Haemophilia can be categorized into three types namely Haemophilia A (factor VIII deficiency), Haemophilia B or Christmas disease (factor IX deficiency), and Haemophilia C or Rosenthal syndrome (low levels of factor XI. However, Haemophilia C differs from Haemophilia A & B in the pattern of bleed tendency and the cause.<sup>2</sup>

Haemophilia, an inherited single X gene disorder arises in the individual through spontaneous /auto mutations or through the known risk of family, gene disorder is assumed

to affect 1per 10,000 birth. The birth prevalence of genetic disorders is likely to be high in India due to due to the high birthrate of 21.8 and the largest birth cohort globally<sup>3</sup>. A meta-analysis study on the incidence of haemophilia recently done indicate that more than 1,125,000 individual across the world is having a bleeding disorder of inheritance. However, in the past, it was considered only around 400,000 only. The expectancy of life for those having haemophilia is significantly less than a normal person, especially in less resource setting areas and countries, where lack of inclusive treatment and rehabilitation leads to chronic joint disorders and organ bleeding result in death and disability. Also, it weakens individual and family, psychologically, and economically.<sup>4,5</sup> As per the 2011 census of India, according to population data, the prevalence of haemophilia A is recorded 4 per 1, 00,000. Nearly 70,000 patients with haemophilia are estimated with 48,407 with Haemophilia A. Intervention of medical and rehabilitation approach is indicated to improve the quality of life of hemophilic to decrease the burden of family and society.<sup>3</sup> Haemophilia is included among the 21 types of disabilities categorized by govt. of India under Rights of Persons

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ISSN: 2231-2196 (Print)

ISSN: 0975-5241 (Online)

Received: 28.08.2020

Revised: 18.10.2020

Accepted: 12.11.2020

Published: 16.02.2021

with Disabilities (RPwD), Act 2016, which gives them equal rights and opportunities as others.<sup>6</sup> The review is aimed to communicate the best clinical orthotic treatment available to hemophilic patients with evidence. Also, focusing on future perspectives and effective approaches for inclusive care to accomplish the quality of life.

## METHODOLOGY

An online literature search was conducted through PubMed, Wiley, Research Gate, Google Scholar, Internet searches, etc using the keywords and headings related to haemophilia and orthotic intervention in the ankle, knee, hip, elbow, etc. The PubMed central mesh word search strategy followed to meet the required literature search. Language restrictions were not applied while searching the articles. These included conference proceedings, organizational reports, newsletters, media stories, and information from websites found via internet searches. Besides, the cited references in the reviewed articles were investigated. All the articles synthesized to extract the line of orthotic treatment in haemophilia and its efficacy.

### Anatomical Joint Structure

Bleeding, Joint disease of arthritis caused by frequent or inadequately treated bleeding into joints, heart disease, and renal disease are common in a cohort of males with haemophilia<sup>7,8</sup>. Spontaneous joint bleeding and repeated hemarthroses cause hemophilic arthropathy with a severe impact on the quality of life and movement. Joint damage through bleeding targets joints, especially the knee, ankle, hip, and elbow, and shoulder due to weight wearing and/or movement required to perform the activities by the concerned limbs<sup>9</sup>. Iron release from joint bleeding induces a chronic inflammation, which is mediated by cytokinins and pro-angiogenic factors lead to articular cartilage damage. However, articular cartilage damage by both the inflammatory and degenerative mechanisms may occur independently. Inflammation in synovial tissue is characterized by hypertrophy, a high degree of neo-angiogenesis, and migration of inflammatory cells known as synovitis.<sup>10-12</sup>

### An orthopaedic manifestation occurs like

- Hemophilic arthropathy may lead to synovitis, cartilage destruction, joint deformity, pseudotumor, articular destruction
- Intramuscular hematoma evident due to swelling, pain, local warmth, and bruising that typically appear in the overlying skin may lead to nerve compression
- Iliacus hematoma can compress the femoral nerve with paresthesias in the L4 destruction
- Epiphyseal overgrowth may lead to LLD (Leg Length Discrepancy) and flexion contracture, valgus deviation caused by asymmetrical growth of the epiphyses, Perthes' disease, osteophytes, and subchondral cysts
- Fracture and pain due to osteopenia<sup>13,14</sup>

In patients with haemophilia, acute haemorrhage is the most frequent, and lack of proper treatment can lead to pathological changes within the joints like pain and arthropathy, which may cause disability.<sup>15</sup>

### Biomechanical Principals

Pain, arthropathy, and muscle bleed can often result in biomechanical deficit and affect normal neuromusculoskeletal function. In the rehabilitation process orthoses play an important role to correct, align, support, or prevent a deformity and restore function<sup>16,17</sup>. Orthoses work basically on the principle of pressure distribution, equilibrium, and lever arm. In pressure principle efficacy achieved through distributing pressure over the greater surface area to achieve minimal pressure per unit area ( $P=F/A$ ), Where F is Force and A is an area of the force application. Equilibrium means the sum of the forces acting on the body through the orthotic device must be equal to zero and more comfortable and effective orthosis can be ensured by the moment arm and sufficient lever arm length<sup>18</sup>.

In conjunction with other modalities orthotic devices are used for the following purpose;

- To prevent post-surgical deformity or deformity in chronic bleeding joints
- To immobilize painful or bleeding joints
- To correct contracture if joint deformity or contracture is there
- To improve joint function maintaining Joint Range of Motion (ROM)
- To provide support to weak muscles or unstable joint<sup>19</sup>

The lower limb is categorized for ambulation, weight support. However, the upper limb is mainly used as a functional unit utilized for prehension and fine motor movement to perform daily activities.<sup>20,21</sup> Orthosis biomechanics is relevant to understanding joint complications, pressures problems on the limb, and altered gait deviation. A combination of forces applies through orthoses to the human body to align the postural structure and improve function.<sup>22</sup> The main aim of orthotic management is to provide enough stability, offload, movement, and support to come closer to a near-normal gait.<sup>23</sup> The investigation from many researchers found that orthotic implication helps reducing pain, abnormal position of body parts along with correction as an external device.<sup>24</sup>

During running and walking foot orthosis users felt reduced knee pain with load transfer from the tibia to the foot during various phases of the gait cycle.<sup>25</sup> In haemophilia, the characteristics of the orthotic device vary as per the condition of the individual joint to be treated based on the need for mobilization or immobilization. Devices are used to manage and prevent the bleeds and its counter effect on the musculoskeletal system. The main goal of orthosis intervention in such cases is to mobilize, immobilize, or offload the affected body

parts to assist in ambulation and gain stability during gait.<sup>26</sup> In modern time orthosis play a crucial role in aiming protection and correction of affected articulation in a hemophilic patient using a variety of orthotic material available worldwide. The knee joint is the most frequently affected due to the complex structure and crucial function ambulation.<sup>27</sup>

## REVIEW OF LITERATURE

Orthotic device treatments to the body or joint to which they are applied vary according to need of implication to the individual in haemophilia to manage or prevent the bleed in the joint and muscle. The lower limb is used in ambulation; however upper limb helps in fine movement, achieving different prehension patterns and purposeful hold of an object. In hemophilic, joints need to be immobilized, stabilize, protect, and assist the affected joint.<sup>28,29</sup>

### Lower limb

In the US haemophilia population, the prevalence of hip dysfunction that affects the ROM of an individual is 16.7%, respectively lower in this cohort for both ankles (45%) and Knee (42%).<sup>30</sup> To favour early, conservative, and smooth mobilization joints need to be rested in a functional position. In a study on animals, joints indicated the weight-bearing on a bleeding joint can damage the cartilage matrix worsted as compared to the non-weight-bearing side.<sup>31</sup> Therefore, it is recommended to use the weight relieving orthosis (KAFO, cane, crutch) to avoid weight-bearing during ambulation in the early days.<sup>32</sup> And with modern technique in orthotic intervention as weight relieving orthosis, which transfers the body weight to the ground with an offload mechanism the hemophilic significantly improve their joint function and management of arthropathy.<sup>33</sup>

Till the coagulation factor availability in a hemophilic patient, the knee joint articulation is at high risk of being affected due to complex nature and an eminent/main joint at risk of trauma or injury due to the high impact of activity or body weight during the activity of daily living. In the recent era of orthoses, intervention plays an effective role in protecting, preventing, and facilitating the desired Range of Motion (ROM) in the treatment of knee hemarthroses caused by chronic synovitis aims at normalizing the Activity of Daily Living (ADL). Quality and nature of Material also play an important role in the treatment process according to the availability of clotting factors.<sup>34</sup>

In patients with haemophilia, the ankle is among the frequent site of arthropathy, and orthotic devices as foot orthosis and footwear can help relieve this pain and achieving near-normal ADL. Lightweight, strong and cushioning materials like carbon fibre and silicone may be used to improve the efficiency of the device to absorb the shear stress and torsional

forces during walking. These orthoses generally provide gait patterns with offload mechanisms or restrict ankle range of motion to minimize the pain. Also, the studies favour the implication of various kinds of orthotic intervention in hemophilic arthropathy joint to improve in relief to pain, stiffness, difficulty, activity limitation, and social and emotional outcomes.<sup>35,36</sup> A significant improvement in activity and reduction in pain found in the study done on the 16 hemophilic patients with a level of 1-5 stage of joint damage in haemophilia A. The foot pain disability measures indicated a reduction in the level of ankle hemarthrosis (bleeds) and improvement in the quality of life in all the patients using functional foot orthosis over 6 weeks period.<sup>37</sup>

Evidence found that ankle joint power generation is higher during footwear walking contrast to barefoot walking in hemophilic patients having severe ankle arthropathy ( $p < 0.001$ ). In the same study a 3D gait data analysis of hemophilic patient without ankle arthropathy ( $n=5$ ) and hemophilic patient with severe hemophilic ankle arthropathy ( $n=17$ ), and a control group ( $n=17$ ), it is found that during footwear walking the ankle joint load was notably reduced in hemophilic patients groups ( $p=0.039$  and  $P=0.002$ ). However, it could not impact much in the control group ( $p=0.952$ ).<sup>38</sup> A study analyzing the rationale of pain and gait pattern using two different braces done. 17 hemophilic patients involved in the study using Fracture Boot (FB) and a Carbon Fiber Floor Reaction Orthosis (CF-FRO). Interestingly both the devices relieved the pain. However, FB changed the gait patterns of the users and CF-FRO did not alter the pattern of gait.<sup>39</sup> Functional Foot Orthoses (FFO) may be implicated to hemophilic patients who suffer valgus or pes planus foot deformity and fixed plantarflexion to reduce the pain and forefoot pressure by shifting load and minimizing shear and translational forces from metatarsal phalangeal joint and controlling forces at subtalar and midtarsal joints.<sup>40</sup>

### Upper limb

Due to the complexity of the human hand and much concurrent joint motion upper limb orthosis management differ from other devices. Special attention is needed on mechanical efficacy, precision to fit with better soft tissue padding for better comfort and acceptance. WHO (Wrist Hand Orthosis), elbow orthosis, and arm sling and be fitted as per need of an individual<sup>41</sup>. In patients with musculoskeletal problems in haemophilia, an orthotic device promotes the quality of life in performing daily activities and sports & games as well.<sup>42</sup>

## FINDINGS

While each participant served their control during studying painful ankle on braces and the unbraced side as well. The pain was relieved significantly ( $p < 0.05$ ) with both devices

compared with the footwear-only (no brace) environment. The difference of pain between fracture boot and carbon fibre AFO(Ankle foot orthosis) was zero that is  $P = 0.999$  during the finding of static effects on bracing it was observed that the swing phase of the braced side was longer to allow the boot to advance which promotes on the unbraced side.<sup>43</sup>

Shoe insoles help in cushioning and modifying stance and prevent lesions. Hemophilic patient with musculoskeletal problems improved their quality of life and restored normal patterns of gait using orthosis and assistive devices. These are also helpful in restoring sports and game activities.<sup>42</sup> Orthotic devices can be used to minimize pain and promote quality life among hemophilic patients controlling joint movement, relieve load, or stabilize a specific joint.<sup>44</sup> Clinical trials on hemophilic favors the improvement in ROM using orthosis. However, there is no difference was found in a randomized control trial between static progressive and dynamic orthosis.<sup>45</sup> The degree of arthropathy increases misalignment. The insole was then frequently used alternative (86%) in hemophilic misalignment to improve patient pain and function on the American Orthopaedic Foot Ankle Society (AOFAS) Ankle-Hindfoot scale.<sup>46</sup>

## PERSPECTIVES

The joints frequently sustain damage in hemophilic conditions and the respective joints should be immobilized and the patient discouraged from walking. Commencement of activity may start after cessation of bleed and improvement in muscle and motion control. The main aim of orthosis in such cases is to offload and immobilize the affected segment and to provide a stable gait pattern during ambulation. Modified insoles and footwear may improve ambulation through absorbing shock, and assistive devices like a wheelchair, walker, cane, crutches, etc. may use to restore normal gait. An orthotic device improves the quality of life in patients with neuromusculoskeletal lesions and is advantageous in preventing deformities from strenuous activities in the course of daily activities.<sup>26</sup> Orthotist can adapt and set the treatment goal as per the clotting factors and extensively available materials available around the world.<sup>47</sup>

## CONCLUSION

The multidisciplinary team approach is highly needed to treat and follow-up hemophilic patients. However, team members should know the roles, responsibilities and their limitations of their own treating such patients, and regular follow-up of every patient is recommended to concern specialist including a physician, orthopedist, coagulation expert, orthopaedic surgeons, etc. to oversee associated problems. Effectively,

a Multidisciplinary haemophilia clinic or institution can play important role in evaluating the condition of patient's joints, their concurrent need, and impacts on the quality of life. While, orthotist play an important role in maintaining and improving their quality of life with optimal activeness through the intervention of appropriate orthosis devices to unload, restrict, or by providing the desired range of motion to the respective affected joints of an individual. In haemophilia chronic synovitis and arthropathies are manageable nowadays with advances in radiologic evaluation and orthotic/orthopaedic interventions. An individual with haemophilia is more likely to have better ADL and functional outcome comparatively than ever before. Though complications like chronic synovitis and arthropathy in haemophilia are manageable nowadays to enjoy better functional results and comfort than ever before. Headway, the advancement in orthotic material, technology, and cost-effective availability to a hemophilic patient shall notably improve musculoskeletal bleeding and related complications. The changes in practice consideration, policy development, and further research development targeting the inclusive well-being of hemophilic health care shall add more rejoice to their lives and dimension in their interaction with society.

In this connection, advanced diagnostic protocols or validated criteria are highly needed to identify the level of haemarthrosis or arthropathy to intervene the orthosis efficiently. Moreover, advancement in materials to improve the comfort level and hygiene of the user, advancement in orthotic fabrication technologies to get the precise shape, and cosmesis for the better user experience and availability of such devices across the urban and rural areas are highly needed. Therefore, further research and study are recommended to obtain better stability and shreds of evidence of the efficacy of orthotic devices treating haemophilia.

## ACKNOWLEDGEMENT

The authors are grateful to Dr. Roshan Bijlee KN, the director of Composite Regional Center for Skill Development, Rehabilitation and Empowerment of Persons with Disabilities, Kozhikode, Kerala, and Mr. Binoy Mathew KV, Lecturer, Physiotherapy at Composite Regional Center for Skill Development, Rehabilitation and Empowerment of Persons with Disabilities, Kozhikode, Kerala for their constant technical support.

**Conflicts of Interest:** No potential conflict of interest was reported by the authors.

**Source of Funding:** Nil

**Authors Contribution:** Authors contributed equally to work conception and design, data collection and analysis, drafting, and critical revision of the article and final approval version to be published.

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