Evidence in Haemophilia Assessment: The Haemophilia Joint Health Score

Lukas Graf[1]

[1] Centre for Laboratory Medicine, Haemostasis and Haemophilia Centre, St. Gallen, Switzerland

Hamostaseologie

Address for correspondence Dr. med. Lukas Graf, Zentrum für Labormedizin St. Gallen, Frohbergstrasse 3, 9001 St. Gallen, Schweiz (e-mail: lukas.graf@zlmsg.ch).

Abstract

Haemophilia is a rare genetic bleeding disorder that primarily affects males and results in the deficiency of clotting factors VIII (haemophilia A) or IX (haemophilia B). One of the most debilitating long-term complications of haemophilia is chronic joint damage with pain, and reduced mobility, due to bleeding into the joints. As the primary cause of morbidity in people with haemophilia (PwH), joint health assessment is critical for disease management and optimizing patient outcomes. Among the tools developed to monitor joint health in PwH, the Haemophilia Joint Health Score (HJHS) has emerged as the most widely used and validated clinical tool. There is evidence supporting the use of the HJHS in both children and adults. In contrast to scoring methods that incorporate imaging techniques, which primarily describe the morphology of the joints, the HJHS allows for the assessment and monitoring of joint functionality.

Keywords
- haemophilia
- joint health
- score

Introduction

Patients with haemophilia A or B are at risk of joint complications, primarily due to recurrent bleeding episodes into the synovial spaces. These bleeding events can result in synovitis, cartilage degradation, and ultimately haemophilic arthropathy.[1] Preventing and managing joint disease is a cornerstone of modern haemophilia treatment, with joint health monitoring critical for clinical decision-making, patient management, and optimizing prophylaxis strategies.[2] Traditionally, clinical assessments of joint function were subjective and lacked standardized, objective tools.[3] The development of the Haemophilia Joint Health Score (HJHS) marked a pivotal shift in the assessment and management of joint health in PwH. This tool provides an objective, systematic evaluation of joint function and damage and has become a key component in clinical trials and routine clinical practice.[4]

Development of the Haemophilia Joint Health Score

The HJHS was first introduced in 2006 as an evolution of earlier joint assessment scales, such as the World Federation of Hemophilia (WFH) physical examination scale.[4,5] The HJHS was designed to address several limitations of previous tools, including their subjectivity and inability to detect early, subtle joint changes. The original HJHS incorporated 12 items across six index joints (elbows, knees, and ankles), assessing joint swelling, muscle atrophy, range of motion, strength, pain, and crepitus.

In 2011, a revised version of the HJHS, HJHS 2.1,[6–8] was released after extensive validation studies. This version refined the scoring system, improved item clarity, and standardized training for healthcare professionals administering the score. It reduced potential inter-rater variability, which was a challenge in earlier versions of joint assessment tools.

In addition to a detailed assessment of the range of motion in the elbows, knees, and ankles, HJHS 2.1 includes explicit definitions and scoring guidelines for joint parameters such as swelling and muscle atrophy. Moreover, HJHS 2.1 also considered age-appropriate scoring, recognizing that joint damage is less frequent in younger children but progresses with age, especially without adequate prophylaxis.[6,7,9] An overview of the Haemophilia Joint Health Score (HJHS) version 2.1 is presented in ►**Table 1**.

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Evidence Supporting the Use of HJHS

Reliability and Validity

Numerous studies have demonstrated the HJHS's reliability and validity in assessing joint health in PwH. A key study by Feldman et al[7] found that the HJHS had excellent inter-rater reliability when administered by trained assessors, highlighting its utility in both clinical trials and routine care settings. Furthermore, the HJHS has been compared with imaging modalities such as magnetic resonance imaging (MRI) and ultrasound. It has been found that clinical (HJHS) and radiological assessment (US/MRI) provide complementary information and should be considered conjointly in the assessment of early joint arthropathy.[10] A 2014 study by Doria et al[11] confirmed that higher HJHS scores were significantly associated with MRI-detected joint damage, reinforcing the score's validity as an indicator of joint health.

Sensitivity to Change

The HJHS has also proven sensitive to detecting changes over time, making it a valuable tool for monitoring the progression of joint disease and the effectiveness of treatment interventions.[12,13] Studies have shown that PwH on prophylaxis tend to have lower HJHS scores compared with those receiving on-demand treatment, reflecting the role of prophylaxis in preventing joint damage.[12,14] Moreover, longitudinal studies have demonstrated that worsening HJHS scores correlate with increased bleeding episodes, underlining the score's sensitivity in capturing disease progression.[15,16]

Use in Paediatric Populations

Early detection of joint changes is critical in paediatric patients with haemophilia, as joint damage is often irreversible once established.[17] The HJHS has originally been developed and validated for use in children as young as 4 years old, with age-adjusted scoring to account for developmental differences.[7] HJHS is reliable in paediatric populations[18] and can detect early signs of joint damage, even in young children on prophylaxis. Detection of these early signs is important because higher HJHS in adolescence and early adulthood correlated with worse joint outcomes later in life.[15]

Use in Adult Populations

Although HJHS has been initially validated for paediatric use, it has been adapted for adults with modifications to address age-related joint changes and degeneration.[6] Its repeated use in adult populations allows for consistent monitoring of joint health and assessment of therapeutic efficacy, thereby supporting individualized treatment planning.[19] However, applying the HJHS to adults requires adjustments that account for age-specific joint pathology and comorbidities. These adaptations are crucial to accurately reflect joint health changes distinct from those observed in younger patients.[6,13]

Integration with Imaging Modalities

Although the HJHS is a valuable clinical tool, its utility is enhanced when combined with imaging modalities like MRI or ultrasound. MRI and ultrasound can detect subclinical joint changes that are not always reflected in the HJHS score, particularly in the early stages of joint disease.[20,21] Combining these assessments provides a more comprehensive understanding of joint health, allowing clinicians to intervene earlier to prevent irreversible damage.[22] A study by Poonnoose et al emphasized the complementary nature of the HJHS and imaging modalities, suggesting that a multimodal approach offers the best strategy for monitoring joint health in PwH.[10] However, unlike MRI or ultrasound, which focus on structural changes, the HJHS provides a functional perspective that is essential for understanding the real-life impact of joint health on movement and daily activities.[7] A comparison

Table 1 Overview of the Haemophilia Joint Health Score (HJHS) 2.1, summarizing joint-specific parameters, scoring ranges, and definitions (The score assesses six index joints and gait, with higher values indicating greater joint impairment)

Joint parameter	Scoring range	Description
Swelling	0–3	0 = None, 1 = Mild (puffy), 2 = Moderate (spongy), 3 = Severe (tense, bony landmarks obscured)
Duration of swelling	0–1	0 = No swelling or <6 months, 1 ≥ 6 months
Muscle atrophy	0–2	0 = None, 1 = Mild (less contour), 2 = Severe (marked wasting)
Crepitus on motion	0–2	0 = None, 1 = Mild (slightly audible/palpable), 2 = Severe (grinding/crunching)
Flexion loss	0–3	0 = < 5 degrees, 1 = 5–10 degrees, 2 = 11–20 degrees, 3 ≥ 20 degrees (worse of contralateral or normative comparison)
Extension loss	0–3	Same criteria as for flexion loss
Joint pain	0–2	0 = None, 1 = Pain on palpation or overpressure, 2 = Pain through active range
Strength	0–4	0 = Normal (gr. 5), 1 = Moderate resistance (gr. 4), … 4 = Trace or none (gr. 1/0)
Global gait	0–4	0 = All skills normal (walk, stairs, run, hop), up to 4 = No skill within normal limits
Assessed joints	–	Bilateral elbows, knees, and ankles (6 joints total)
Maximum score	**124**	**Total sum of joint items and gait (if all joints evaluable)**

Table 2 Differences between the Haemophilia Joint Health Score (HJHS) and imaging technologies (like MRI and ultrasound) in assessing joint health in haemophilia patients

Criteria	Haemophilia Joint Health Score (HJHS)	Imaging technologies (MRI, ultrasound)
Purpose	Clinical assessment of joint function and physical signs	Detection of structural and subclinical joint changes
Sensitivity	Limited in detecting early, subclinical changes	Highly sensitive, detects early and subtle joint damage
Assessed joints	Large joints (elbows, knees, ankles)	Can assess large and small joints (including wrists, shoulders, etc.)
Cost and accessibility	Low-cost, requires only trained healthcare professionals	Higher cost, requires specialized equipment and expertise
Time required	Relatively quick (few minutes per joint)	Time-consuming (especially MRI, which can take 30+ minutes per joint)
Training and expertise	Requires training for consistency but relatively straightforward	Requires radiologists (MRI) and specialized training
Intervention focus	Focuses on visible or palpable joint damage and symptoms	Focusses on detailed internal joint changes and structure
Utility in disease progression	Monitors clinical changes over time, useful in routine care	Monitors disease progression at a deeper, structural level
Limitations	Subjective; may miss subclinical changes	Expensive, limited availability in some clinical settings

of joint assessments using HJHS and imaging technologies is presented in ►Table 2.

Limitations of the HJHS

Although the HJHS is a widely used tool, it has certain limitations. One of the key challenges is its relative insensitivity to very early, subclinical joint changes that may be detected by advanced imaging techniques.[23,24] Although MRI and ultrasound are not always feasible in every clinical setting, they are crucial in detecting the earliest signs of joint damage, which the HJHS may miss.[25]

Another limitation is the need for trained assessors to ensure consistent and accurate scoring.[26] Although training programs exist to standardize HJHS administration, there can still be variability in its application across different centres and healthcare providers.

Furthermore, the HJHS focuses on large joints (elbows, knees, and ankles) and does not assess smaller joints like wrists or shoulders, which may also be affected by haemophilia-related joint disease.[6] This limits the comprehensiveness of the score in evaluating overall joint health in some patients.[27]

The applicability of the HJHS in patients with advanced joint disease may be limited. In individuals who have undergone joint arthroplasty or arthrodesis, several domains—particularly range of motion, muscle strength, and crepitus—cannot be reliably assessed, resulting in 'non-evaluable' entries. These gaps reduce the interpretability of the total score and complicate longitudinal assessments or comparisons between patients.[6,27,28] Moreover, the HJHS may underestimate joint impairment in end-stage arthropathy, where joints are structurally damaged but clinically silent, yielding disproportionately low scores compared with joints affected by active synovitis. As a result, patients with 'quiet' yet severely damaged joints may appear to have better joint health than is actually the case.[28,29]

The Role of the HJHS in the Future of Haemophilia Care

As haemophilia treatment continues to advance with increasingly effective therapies, the ultimate goal is to provide individuals with haemophilia a life free from bleeding episodes.[30] Tools like the HJHS will continue to play a crucial role in monitoring joint health. Remote monitoring through digital health tools and further refinement of the HJHS, including patient-reported outcomes, could enhance its utility in future haemophilia care.[31]

Conclusion

The HJHS has become an invaluable tool for assessing and monitoring joint health in PwH. Its reliability, validity, and sensitivity to change have made it a cornerstone of clinical practice and research. Despite its limitations, the HJHS provides a standardized, objective measure of joint damage, which is critical for optimizing prophylaxis, tracking disease progression, and improving patient outcomes. While imaging technologies provide detailed structural information, the HJHS focusses on functional implications of joint damage, making it more aligned with patient-reported outcomes and quality of life.

As the field of haemophilia care continues to evolve, the HJHS will remain a useful tool in ensuring that joint health is appropriately managed and protected in PwH.

Conflict of Interest

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