Surveillance de la santé articulaire et impact de la rééducation dans l'hémophilie pédiatrique : expérience d'un centre unique

Joint Health Monitoring and the Impact of Rehabilitation in Pediatric Hemophilia: A Single-Center Experience

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RÉSUMÉ

Introduction : L'arthropathie hémophilique constitue une cause majeure de morbidité chez l'enfant atteint d'hémophilie sévère. Les hémarthroses répétées entraînent une synovite chronique, responsable de lésions articulaires progressives et d'un retentissement fonctionnel important. Une surveillance précoce associée à une rééducation adaptée pourrait limiter l'évolution vers le handicap.

Objectif : Évaluer l'évolution de la santé articulaire et l'impact de la rééducation fonctionnelle chez des patients pédiatriques atteints d'hémophilie sévère.

Méthodes : Une cohorte de 101 enfants atteints d'hémophilie sévère a été suivie. Seize patients ont développé une arthropathie hémophilique intéressant 24 articulations. L'évaluation reposait sur le score clinique HJHS, le score radiologique de Pettersson et l'IRM. Les patients ont été évalués avant et après mise sous prophylaxie et rééducation fonctionnelle. L'évolution du HJHS a été analysée aux niveaux global (ΔHJHS-total) et articulaire (ΔHJHS-joint). Les comparaisons avant/après ont été réalisées à l'aide du test des rangs signés de Wilcoxon, avec un seuil de significativité fixé à $p < 0,05$.

Résultats : L'âge médian était de 12 ans [6-17], et trois patients présentaient des inhibiteurs. Les genoux étaient les articulations les plus atteintes (75 %), suivis des coudes et des chevilles (12,5 % chacune). Le HJHS total médian initial était de 32. Après rééducation, une diminution statistiquement significative du HJHS-total a été observée (réduction médiane : −13 points ; $p < 0,001$). Une amélioration de la douleur, de l'amplitude articulaire, de la force musculaire et de la tolérance à la marche a été constatée. Les scores de Pettersson sont restés stables ($p > 0,05$), tandis que l'IRM montrait une stabilisation ou une régression partielle de la synovite, sans modification structurale significative.

Conclusion : La surveillance articulaire précoce associée à une rééducation fonctionnelle structurée est associée à une amélioration fonctionnelle significative de l'arthropathie hémophilique pédiatrique. Le HJHS constitue un outil sensible pour le suivi clinique en pratique courante.

Mots clés : Hémophilie sévère ; Arthropathie hémophilique ; Hemophilia Joint Health Score (HJHS) ; Rééducation fonctionnelle ; Imagerie par résonance magnétique (IRM).

ABSTRACT

Background: Hemophilic arthropathy is a major cause of morbidity in children with severe hemophilia. Recurrent hemarthroses lead to progressive joint damage, chronic synovitis, and functional impairment. Early joint monitoring combined with structured rehabilitation may limit long-term disability.

Objective: To evaluate the evolution of joint health and to assess the impact of functional rehabilitation in pediatric patients with severe hemophilia.

Methods:

A cohort of 101 children with severe hemophilia was followed at our center. Sixteen patients developed hemophilic arthropathy involving 24 joints. Joint status was assessed using the Hemophilia Joint Health Score (HJHS), the Pettersson radiological score, and magnetic resonance imaging (MRI). Evaluations were performed before and after prophylaxis and functional rehabilitation. Changes in HJHS were analyzed at both patient (ΔHJHS-total) and joint (ΔHJHS-joint) levels. Pre- and post-rehabilitation comparisons were performed using the Wilcoxon signed-rank test, with statistical significance set at $p < 0.05$.

Results: The median age was 12 years [6-17], and three patients had inhibitors. Knees were the most frequently affected joints (75%), followed by elbows and ankles (12.5% each). The median baseline total HJHS was 32. After rehabilitation, a statistically significant reduction in HJHS-total was observed (median decrease: −13 points; $p < 0.001$). Improvements were noted in pain, range of motion, muscle strength, and walking tolerance. Pettersson scores remained stable ($p > 0.05$), while MRI findings showed stabilization or partial regression of synovitis, without statistically significant structural changes.

Conclusion: Early joint monitoring combined with structured functional rehabilitation is associated with significant functional improvement in pediatric hemophilic arthropathy. The HJHS appears to be a sensitive tool for assessing clinical outcomes in routine practice.

Keywords: Severe hemophilia; Hemophilic arthropathy; Hemophilia Joint Health Score (HJHS); Functional rehabilitation; Magnetic resonance imaging (MRI).

INTRODUCTION

Joint bleeding in patients with hemophilia can lead to progressive hemophilic arthropathy. Regular monitoring of joint health is essential to detect early deterioration and to allow timely therapeutic and rehabilitative interventions.

OBJECTIVE

To evaluate the evolution of joint health in children with moderate to severe hemophilia and to assess the contribution of functional rehabilitation in preserving joint function.

METHODS

A retrospective cohort study was conducted in children with moderate and severe hemophilia followed at our center. Joint health of the ankles, knees, and elbows was assessed using the Hemophilia Joint Health Score (HJHS).

Analyses were performed at:

- the patient level (ΔHJHS-total), calculated as the sum of affected joint scores, and
- the joint level (ΔHJHS-joint).

Joint deterioration was defined as ΔHJHS-total ≥ 4 at the patient level and ΔHJHS-joint ≥ 2 at the joint level.

Radiological damage was evaluated using the Pettersson score. MRI assessment was performed when available to characterize synovial and osteochondral involvement.

STATISTICAL ANALYSIS

Inclusion :

Quantitative variables were expressed as median and interquartile range (IQR). Pre- and post-rehabilitation comparisons were performed using the Wilcoxon signed-rank test due to the small sample size and non-normal distribution of variables. A p-value < 0.05 was considered statistically significant

RESULTS

A total of 101 children with severe hemophilia were followed at our center. Among them, 16 patients developed hemophilic arthropathy, corresponding to 24 affected joints.

The median age was 12 years [6-17], and three patients had inhibitors.

At baseline, the median HJHS per joint was 14 [6.25], and the median Pettersson score was 8 [4-12], indicating established structural joint damage.

Joint involvement predominantly affected the knees (75%, n = 18), followed by the elbows (12.5%, n = 3) and ankles (12.5%, n = 3) (Figure 2). Two patients presented with involvement of three joints, while four patients had two affected joints.

Baseline clinical, radiological, and MRI characteristics are summarized in Table 1. Before rehabilitation, HJHS values reflected moderate to severe functional impairment, particularly in knee joints. Pettersson scores confirmed chronic structural damage, while MRI findings showed variable degrees of synovitis and osteochondral involvement.

Regarding therapeutic management, seven patients demonstrated clinical improvement after combined prophylactic treatment and functional rehabilitation, while three patients improved following synoviorthesis. Five newly diagnosed patients were initiated on prophylaxis and functional rehabilitation, with follow-up ongoing.

After rehabilitation, a statistically significant reduction in HJHS-total was observed. The median total HJHS decreased from 32 at baseline to 19 after rehabilitation, corresponding to a median reduction of −13 points (Wilcoxon signed-rank test, $p < 0.001$).

At the joint level, ΔHJHS-joint values were significantly negative across affected joints ($p < 0.001$), confirming objective improvement in joint function.

In contrast, Pettersson scores remained globally stable after rehabilitation, with no statistically significant difference between baseline and follow-up values ($p > 0.05$), consistent with the irreversible nature of structural joint damage.

MRI findings demonstrated stabilization or partial regression of synovitis in several joints; however, these changes did not reach statistical significance.

The evolution of joint health is detailed in Table 3. All patients showed functional improvement, with decreases in HJHS-total ranging from −2 to −32 points. The most pronounced improvements were observed in patients who benefited from combined prophylaxis, intensive functional rehabilitation, and synoviorthesis.

Fig. 1 | Hemophilia Joint Health Score (HJHS)

Subject ID #:_____ Name of Physiotherapist:_____

Assessment #: _____ Date: _____
Time: _____ yyyy / mm / dd

Hemophilia Joint Health Score 2.1 - Summary Score Sheet

	Left Elbow	Right Elbow	Left Knee	Right Knee	Left Ankle	Right Ankle
Swelling	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE
Duration (swelling)	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE
Muscle Atrophy	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE
Crepitus on motion	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE
Flexion Loss	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE
Extension Loss	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE
Joint Pain	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE
Strength	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE	☐ NE
Joint Total						

Sum of Joint Totals + ☐
Global Gait Score ☐
(☐ NE included in Gait items)
HJHS Total Score = ☐

NE = Non-Evaluable

Swelling
0 = No swelling
1 = Mild
2 = Moderate
3 = Severe

Duration
0 = No swelling or < 6 months
1 = ≥ 6 months

Muscle Atrophy
0 = None
1 = Mild
2 = Severe

Crepitus on Motion
0 = None
1 = Mild
2 = Severe

Flexion Loss

	Contralateral:	Normative Tables:
0 =	< 5°	0= within range
1 =	5° - 10°	1 = 1° - 4°
2 =	11° - 20°	2 = 5° - 13°
3 =	> 20°	3 = > 10°

Extension loss (from hyperextension)

	Contralateral:	Normative tables:
0 =	< 5°	0= within range
1 =	5° - 10°	1 = 1° - 4°
2 =	11°- 20°	2 = 5° - 13°
3 =	> 20°	3 = > 10°

Joint Pain
0 = No pain through active range of motion
1 = No pain through active range; only pain on gentle overpressure or palpation
2 = Pain through active range

Strength (Using The Daniels & Worthingham's scale)
Within available ROM
0 = Holds test position against gravity with maximum resistance (gr.5)
1 = Holds test position against gravity with moderate resistance (but breaks with maximal resistance) (gr.4)
2 = Holds test position with minimal resistance (gr. 3+) or holds test position against gravity (gr.3)
3 = Able to partially complete ROM against gravity (gr.3-/2+), or able to move through ROM gravity eliminated (gr.2), or through partial ROM gravity eliminated (gr.2-)
4 = Trace (gr.1) or no muscle contraction (gr.0)
NE = Non-evaluable

Global Gait (walking, stairs, running, hopping on 1 leg)
0 = All skills are within normal limits
1 = One skill is not within normal limits
2 = Two skills are not within normal limits
3 = Three skills are not within normal limits
4 = No skills are within normal limits
NE = Non-evaluable

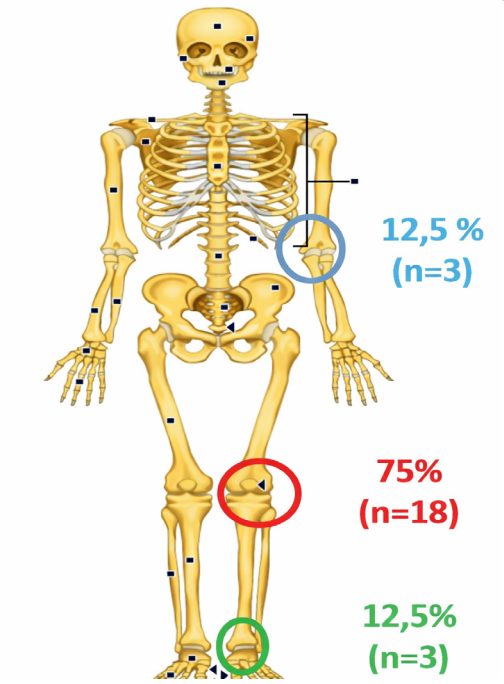
NOTE: There is an accompanying instruction manual and worksheets that are required when administering the HJHS

General Comments:

_____
_____
_____
_____

Hemophilia Joint Health Score 2.1 , © The Hospital for Sick Children, Centre Hospitalier Universitaire Sainte Justine, the Regents of the University of Colorado, Karolinska Hospital, University Medical Center Utrecht, 2009. Used under license by The Hospital for Sick Children

Fig. 2 | Distribution of arthropathies



Tab. 1 | Characteristic hemophilic arthropathy before rehabilitation

Case	Age (y)	Joint(s)	HJHS	Pettersson	MRI score
1	15	L. Ankle	6	4	3
2	7	R. Ankle	6	4	5
3	13	R. Knee / L. Knee	17 / 17	10 / 10	12 / 12
4	8	R. Knee / L. Knee	25 / 21	11 / 11	16 / 14
5	17	R. Knee / L. Knee / R. Elbow	25 / 18 / 14	12 / 12 / 8	17 / 12 / 8
6	6	R. Knee / L. Knee / R. Ankle	14/ 18 / 10	8/ 12 / 6	10/10/6
7	9	L. Knee	12	6	8
8	11	R. Knee	10	6	7
9	14	L. Knee	16	8	11
10	12	R. Knee / L. Knee	18/18	9/8	13/12
11	16	L. Knee	20	11	15
12	10	R. Knee	13	7	9
13	15	L. Knee	22	12	16
14	9	R. Elbow	11	6	7
15	13	L. Elbow	15	8	10
16	17	R. Knee / L. Knee	19 / 16	10 / 9	14 / 12

Tab. 2 | Characteristic hemophilic arthropathy after rehabilitation

c	Age (y)	Joint(s)	HJHS	Pettersson	MRI score
1	15	L. Ankle	1	0	-
2	7	R. Ankle	5	3	ND
3	13	R. Knee / L. Knee	10 / 10	10 / 10	- / -
4	8	R. Knee / L. Knee	11 / 8	11 / 11	15 / 15
5	17	R. Knee / L. Knee / R. Elbow	18 / 12 / 10	12 / 12 / 12	9 / 8 / 8
6	6	R. Knee / L. Knee / R. Ankle	12/12/6	6/8/6	ND
7	9	L. Knee	8	6	7
8	11	R. Knee	9	7	8
9	14	L. Knee	12	8	11
10	12	R. Knee / L. Knee	14/6	9/7	12
11	16	L. Knee	15	11	14
12	10	R. Knee	10	7	8
13	15	L. Knee	16	12	15
14	9	R. Elbow	8	6	6
15	13	L. Elbow	11	8	9
16	17	R. Knee / L. Knee	14 / 12	10 / 9	13 / 11

Tab. 3 | ΔHJHS-total and ΔHJHS-joint

c	Age (y)	Joint(s)	HJHS	Pettersson	MRI score
1	15	L. Ankle	1	0	–
2	7	R. Ankle	5	3	ND
3	13	R. Knee / L. Knee	10 / 10	10 / 10	– / –
4	8	R. Knee / L. Knee	11 / 8	11 / 11	15 / 15
5	17	R. Knee / L. Knee / R. Elbow	18 / 12 / 10	12 / 12 / 12	9 / 8 / 8
6	6	R. Knee / L. Knee / R. Ankle	12/12/6	6/8/6	ND
7	9	L. Knee	8	6	7
8	11	R. Knee	9	7	8
9	14	L. Knee	12	8	11
10	12	R. Knee / L. Knee	14/6	9/7	12
11	16	L. Knee	15	11	14
12	10	R. Knee	10	7	8
13	15	L. Knee	16	12	15
14	9	R. Elbow	8	6	6
15	13	L. Elbow	11	8	9
16	17	R. Knee / L. Knee	14 / 12	10 / 9	13 / 11

DISCUSSION

Chronic hemophilic arthropathy remains one of the leading causes of morbidity in patients with hemophilia, despite major advances in clotting factor replacement therapy and the introduction of prophylactic regimens [1-3]. Recurrent joint bleeding episodes result in the accumulation of blood within the synovial cavity, triggering inflammatory cascades that promote synovial hypertrophy, angiogenesis, and progressive cartilage and bone destruction [4, 5]. Over time, this leads to chronic hemophilic synovitis and establishes a self-perpetuating vicious cycle of hemarthrosis–synovitis–hemarthrosis, accelerating joint deterioration and functional impairment [6].

The clinical consequences of hemophilic arthropathy are substantial. Patients frequently experience chronic pain, joint stiffness, muscle weakness, reduced range of motion, impaired gait, and limitations in daily activities [7-9]. These impairments significantly affect quality of life, social participation, and long-term independence [10, 11]. The progression of joint damage typically occurs over 5 to 10 years, particularly in weight-bearing joints such as the knees and ankles [6, 12]. Consistent with previous studies, our results confirm that the knees and ankles were the most frequently affected joints [7, 13].

Joint involvement was mainly localized to the knees, ankles, and elbows, reflecting the typical distribution of hemophilic arthropathy described in the literature [6, 13, 14]. Knee involvement is particularly relevant due to its impact on gait, balance, and overall mobility [15]. Ankle involvement compromises postural stability and walking efficiency, while elbow involvement limits upper-limb function and daily activities [14]

Clinical assessment using the Hemophilia Joint Health Score (HJHS) demonstrated significant improvement in joint function [16, 17]. The total reduction of 131 points reflects meaningful clinical progress in joint mobility, muscle strength, pain reduction, and functional performance [1, 18]. The most pronounced improvements were observed in the knees, likely due to rehabilitation programs emphasizing lower-limb strengthening, gait training, and functional exercises [1, 19].

The HJHS is a validated and sensitive tool for detecting early and moderate joint impairment in hemophilia [16, 17]. Improvements in HJHS scores reflect enhanced functional capacity rather than structural recovery [18]. These findings strongly support the effectiveness of physiotherapy in patients with hemophilic arthropathy [1, 19, 20].

In contrast, radiological assessments using the Pettersson score and MRI showed only slight improvement or stability [21, 23]. This discrepancy between clinical and imaging findings is well documented and reflects different timelines of functional versus structural change [6, 22]. While physiotherapy improves neuromuscular control, muscle strength, and joint stability, structural joint damage often persists [5, 24].

The Pettersson score evaluates chronic radiographic changes such as joint space narrowing, erosions, and deformities, which reflect largely irreversible damage [21]. MRI provides more sensitive detection of synovial hypertrophy, cartilage damage, and hemosiderin deposition [22, 23]. Structural improvements on MRI usually occur slowly, especially in advanced arthropathy [23].

Therefore, imaging stability should not be interpreted as treatment failure but rather as a reflection of the chronic nature of hemophilic joint disease [6, 24]. Clinical improvements remain highly valuable, as they translate into better function and improved quality of life [10, 18].

Physiotherapy plays a central role in the multidisciplinary management of hemophilic arthropathy [1, 19, 20]. Rehabilitation aims to restore joint mobility, improve muscle strength, enhance proprioception, optimize gait, and prevent secondary musculoskeletal complications [15, 20]. Individualized exercise programs are essential, as patients differ in disease severity, joint involvement, age, and functional capacity [18].

Functional and dynamic exercises—including closed-chain strengthening, balance training, gait re-education, and task-specific activities—are particularly beneficial [1, 15, 19] and may reduce joint stress and the risk of future bleeding episodes [5, 24].

Physiotherapy also provides psychological and social benefits. Improved mobility and independence can enhance self-confidence, reduce fear of movement, and encourage greater participation in school, work, and social activities [10, 11].

Early initiation of physiotherapy is crucial to prevent muscle atrophy, maintain joint range of motion, and slow arthropathy progression [6, 20, 24]. Regular monitoring using validated tools such as the HJHS, Pettersson score, and MRI allows early detection of joint changes and supports individualized management [17, 21-23].

Despite limitations such as small sample size and incomplete MRI follow-up, the consistent improvement in HJHS scores strongly supports the clinical effectiveness of physiotherapy [18, 20]. The stability of radiological scores suggests prevention of further joint deterioration [23, 25].

Overall, hemophilic arthropathy is both a structural and functional condition. While structural damage may persist, functional recovery through targeted rehabilitation remains achievable and clinically meaningful [18, 24, 25]. Physiotherapy should therefore be considered a standard component of comprehensive hemophilia care [1,3, 20].

CONCLUSION

Chronic hemophilic arthropathy remains a major source of disability in patients with hemophilia. Early detection of joint deterioration through systematic monitoring, combined with structured and continuous physiotherapy, is essential to limit functional impairment. A multidisciplinary approach involving hematologists, physiotherapists, and rehabilitation specialists is crucial to optimize long-term joint outcomes and quality of life in pediatric patients with hemophilia.

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