View metadata, citation and similar papers at core.ac.uk
brought to you by CORE
provided by AJOL - African Journals Online

Original Article

Patterns of Orthopaedic Complications of Haemophilia at Khartoum Haemophilia Clinic.

Shaheen S[1] , Gashi Y[2] and Satti M[3]

Abstract:

Background: Haemophilia is a common hereditary bleeding disorder caused by deficiency in clotting factor VIII (Type A) factor IX (Type B) with A:B 5:1. Severity of the disease depends on the level of the circulating factor. Bleeding tendency is the presenting feature and musculoskeletal involvement is a common presenting feature.

Objectives: To study the demographic characteristics, clinical and radiological patterns of musculoskeletal disorders associated with haemophilia in patients presented to Khartoum Haemophilia Clinic (KHC).

Patients and Methods: Demographic characteristics, patterns of clinical and radiological features of 78 patients with haemophilia A and B who presented to KHC, between March 2004 and June 2005 were analyzed.

Results: There were 78 patients; all were males, their ages ranging between 1.5 and 50 years. 80% of them were either of preschool or school age groups. Haemophilia A: B was 4.5:1.

Over 80% had articular involvement and the knee joint was involved in more than 50% of cases. Radiological findings were less severe in patients with haemophilia B, and were more severe in patients older than 30 years of age.

Conclusion: We conclude that most of patients present with sequelae of recurrent musculoskeletal bleeds. Thus we observed that most of cases presented late with already destroyed joints. We recommend here to give treatment as prophylactic rather than on demand as it is now practiced as inevitable destruction of joints with repeated bleeds will be the presenting feature.

Keywords: Musculoskeletal, hyperaemia, pseudotumours.

Haemophilia is a common hereditary bleeding disorder that occurs due to deficiency in clotting factors VIII (Type A) and IX(Type B). It is an X-linked recessive bleeding disorder; however may occur sporadically due to mutation and very rarely acquired following an immune abnormality[1]. Incidence of haemophilia A is 1 in 5000 – 10000 and that of Haemophilia B is 1 in 25000, and no racial difference is observed[2, 3]. The main abnormality is deficiency of the factor in 90% of cases, but the cause is defective factor in the rest (10%).

1. Department of Orthopaedics and Traumatology. Faculty of Medicine University of Khartoum. Sudan.
2. Department of Pathology. Faculty of Medicine University of Khartoum, Sudan.
Correspondence: Assoc. Prof. Samir Shaheen.
Email: drsshaheen@hotmail.com

There is no difference in clinical presentation between both types but severity is directly related to the level of circulating factors[4, 5] and according to this level it is classified to severe, moderate and mild[6]. There are no differences in radiological findings between both types[7]

In severe cases patients may present with spontaneous bleeding and 80% of bleeding targets knee, ankle and elbow joints[8], therefor; haemoarthosis is the most common orthopaedic problem and can occur as early as two years of age[9-11]. Asymmetrical epiphyseal growth results from recurrent bleeds which causes inflammatory reaction and hyperaemia[8].

Common presenting features in haemophilic patients are limitation of movement, fixed deformities, wasting and sometimes joint

instability. Pain is not a striking feature, but joint cartilage involvement leads to joint destruction[12].

Intramuscular bleeding occurs in more than 10% of orthopaedic complications, psoas, forearm and calf muscles being the most commonly affected[11]. Haemophilic pseudotumours are rare but serious complication[13].

Objective:

To study the demographic characteristics as well as clinical and radiological patterns of musculoskeletal disorder associated with haemophilia in patients presented to Khartoum Haemophilia Clinic (KHC).

Patients and Methods

The study included 78 patients with haemophilia A and B. They presented to KCH between March 2004 and June 2005, from those who presented to the clinic only those who had orthopaedic complications were included. Khartoum Teaching Hospital authorities were informed and all patients or guardians were informed and consented.

All included patients were examined to assess musculoskeletal involvement, radiological examination was also done to the affected site and joint involvement was classified according to Arnold and Hilgartner classification[14].

Data were filled in a standardized form designed to include personal characteristics; name, gender, age, school/work performance. History concerning recent and remote orthopaedic problem was included in addition to the findings of clinical examination and radiological assessment.

Results:

All the patients (78) were males, their ages ranged between 1.5 and 50 years with a mean age of 17.23 years.(table 1).

Among 78 patients 64 (82.1%) have haemophilia A and 14 (17.9%) have haemophilia B.

Majority [67.9% (53 of the 78)] of patients were students, 12.8% (10 out of 78) were below school age, 19.3% (15 out of 78) were labours, employees and unidentified jobs.

Table1: Age group distribution for 78 haemophilic patients with musculoskeletal involvement.

Age (in years)	No. of patients	%
1 – 5	9	11.5
6 – 10	18	23.1
11 – 15	10	12.8
16 – 20	14	17.9
21 – 25	14	17.9
26 – 30	6	7.7
31 – 35	1	1.3
36 – 40	3	3.8
41 – 45	1	1.3
46 – 50	2	2.6
Total	78	100

Most [65 out of 78(83.3%)] of patients, had joint involvement, whereas muscles were affected in 11 (14.1%) patients. Bone was involved in two patients and in another two, both joint and bone were affected, both constituted 5.2%.

More than one joint were affected in 70.5% of patients (n=47). The knee joint constituted 51.3%, being the most commonly affected joint (Table 2).

Table 2: Site of the bleeding on presentation for 78 haemophilic patients

The Site	No.	%
Knee joint	40	51.3
Elbow Joint	11	14.1
Ankle joint	4	5.1
Shoulder joint	7	9.0
Hip joint	1	1.3
Wrist joint	1	1.3
Foot	1	1.3
Hand	2	2.6
Iliopsoas	2	2.6
Calf Muscles	3	3.8
Forearm Muscles	2	1.6
Others	4	5.2
Total	78	100

Swelling, pain with or without movement and decrease in range of motion were the most common presenting complaints (table 3)

Table3: The main presenting symptom/s for 78 haemophilic patients.

symptoms	No	%
Swelling	70	89.7
Pain	69	88.5
Deformity	22	28.2
Pain with movement	66	84.6
Muscle Weakness	37	47.4
Decrease range of movement	52	66.7
Numbness	15	19.2
Others	5	6.4

Onclinical examination, swelling was found in more than 90% of patients (n=72) and tenderness in more than 75% of patients (n=61). Diminished range of movement was found in more than 70% of patients (n=59) (Table 4).

Table3: Clinical findings of presentation 78 haemophilic patients.

The signs	No	%
Swelling	72	92.2
Wasting	36	46.2
Bruises	3	3.8
Deformity	26	33.3
Hotness	35	44.9
Tenderness	61	78.2
Decrease motion	59	75.6
Parasthesia	6	7.7
Compartment syndrome	1	1.3
Crepitus	31	39.7
Limb discrepancy	35	44.9

In 85% of patients who were less than 30 years, radiological findings were stage I, II or III, whereas in those who were older than 30 years 82% of patients had stage IV and V radiological findings. Radiological findings were less severe in patients with haemophilia B than with A.

Poly-articular involvement was also associated with severe radiological features.
Radiological findings in the 67 patients with joint affection, varied from local osteoporosis in 59% (n=46), to narrowing of joint space in 48% (n=38) (Table 5).

Table5: Radiological findings in 78 haemophilic patients.

Radiological findings	No	%
Soft Tissue Swelling	45	57.7
Local Osteoporosis	46	59.0
Epiphysial overgrowth	30	38.5
Decrease joint Space	38	48.7
Subarticular bone cyst	17	21.8
Osteophytes	20	25.6
Pseudotumour	1	1.3
Fracture	2	2.6
Sclerosis (periarticular)	12	15.4

Discussion

Haemophilia is a coagulation disorder sex linked thus it affects males only[15]. Majority of our patients were from the northern and western parts of Sudan, as there is no known racial difference in the incidence of haemophilia as reported by Soucie et.al[3], this clustering of patients in these areas could well be due to easy accessibility from these areas to the only multidisciplinary haemophilia clinic in Sudan. A similar observation was found by Kim, Yang and Lee in Korea[16] who found that more than 40% of their series to have been clustered in one hospital, though they have more than 15 centres compared to only one centre we have in Sudan. Kar and Potnis -Lele[17], found that cases are clustered in areas where haemophilia clinics are present. However some authors reported incidence of haemophilia among Chinese to be one fourth less than among whites[18].
In this study more than 50% of patients were below 16 years of age and this finding goes in line with what reported by Kim et al. This could be explained by that fact that frequency of bleeds decrease with skeletal maturity[8]. Soucie et al, reported that the age specific prevalence of haemophilia declines steadily with age; being the highest in the age group 4-15 years[3]. Haemophilia A is commoner than B; and in this study it constituted more than 80 % of cases. Similar results were also reported by others[16,19, 20].
Kim and Manisha[16, 19], found in their series, that work performance among haemophilic patients was poor, a finding which also could

be observed in this study with more than 25 % of patients having poor work performance as students or as workers. This could well be due to multiple off days from work in repeated attacks.

Haemophilic arthropathy once occurred undergoes progressive course with more damage after each bleed[8]. Appoint to be mentioned here is that, the use of prophylactic factor VIII reduces the development of haemathroses and haemophilic arthropathy[14], however how long a joint needs to be permanently damage is still unclear[8].

Many authors[7, 9, 12, 21], found that joints' bleeds are much common than other musculoskeletal bleeds. In our series joints bleeds constituted more than 80%; and the knee joint was involved in more than 50% of cases. A similar observation was also made by other authors[7, 9, 22]. In earlier studies shoulder joint was found to be the least joint involved in haemophilic bleeds. It was reported to be 0.8% by Kim[16] and 0.2% by Sajid[23]. In this study shoulder joint bleeds constituted 9% of involved joints. Further study is needed to explain the cause of this variation.

Patients presenting with fractures constituted less than 2% of our series, a finding which could be due to the fact that haemophilic patients live a more low profile activity life style and are less prone to trauma as stated by File et al and Biggs[24, 25].

In more than 70% of our patients more than one joint was involved in the same patient. Thomas[26], reported that lower levels of circulating clotting factor is associated with bleeds in more than one joint.

Swelling was a shared presenting feature in more than 90% of cases. Swelling was reported to be mainly due to haemarthrosis[7-9]. Another cause could be the thickened synovium[8].

Radiological changes in haemophilic arthropathy include; osteoporosis, osteonecrosis, epiphyseal overgrowth, bone cyst, narrowing of joint space, soft tissue swelling and bony fusion, however, conventional radiology is unable to show joint effusion and synovial hyperplasia[14].

Many authors reported that there is no difference between haemophilia A and B in radiological features[27], also it was reported that haemophilia B patients are prone to loss of bone density similar to those of haemophilia A[28]. However we found that haemophilia A patients shows more radiological affection when compared to haemophilia B patients. This could be due to the fact that haemophilia A shows more severe course than haemophilia B, even other severe complications of haemophilia were found to be more in A than B[16].

In our findings the intensity of radiological involvement increases with age, a finding that goes in line with earlier reports[9].

From our findings we can say that the severity of radiological changes is also affected by the number of joints affected; patients with poly articular involvement showed more severe radiological involvement than mono-articular patients. This point goes with the fact that there are other reasons for severity of haemophilic arthropathy apart from the level of circulating clotting factor[9].

Conclusion and Recommendations:

This is the first study about orthopaedic complications of haemophilia to be carried out in Sudan. From what we observed, most of cases presented late with already destroyed joints. We recommend here to give treatment as prophylactic rather on demand as it is now practiced as inevitable destruction of joints with repeated bleeds will be the presenting feature.

References:

1. Green D, Lechner K. A survey of 215 non-hemophilic patients with inhibitors to Factor VIII. Thromb Haemost. 1981;45(3):200-3.

2. Biggs R, Douglas AS, Macfarlane RG et al. Christmas disease: a condition previously mistaken for haemophilia. Br Med J. 1952;2(4799):1378-82.

3. Soucie JM, Evatt B, Jackson D. Occurrence of hemophilia in the United States. The Hemophilia Surveillance System Project Investigators. Am J Hematol. 1998;59(4):288-94.

4. Roberts HR, Monroe DM, Oliver JA et al. Newer concepts of blood coagulation. Haemophilia. 1998;4(4):331-4.

5. White B, Lee CA. Diagnosis and management of Inherited Bleeding Disorders. In: Rodriguez-Merchan E C GNj, Lee C A, editor. Muscloskeletal Aspect of Haemophilia. 1st ed. Oxford: Weily-Blackwell; 2000. p. 3-10.
6. Roberts HR. Molecular biology of hemophilia B. Thromb Haemost. 1993;70(1):1-9.
7. Rodriguez-Merchan EC. Common orthopaedic problems in haemophilia. Haemophilia. 1999;5 Suppl 1:53-60.
8. Mulder K, Llinas A. The target joint. Haemophilia. 2004;10 Suppl 4:152-6.
9. Klukowska A, Czyrny Z, Laguna P et al. Correlation between clinical, radiological and ultrasonographical image of knee joints in children with haemophilia. Haemophilia. 2001;7(3):286-92.
10. Rodriguez -Merchan EC, Goddard N. Muscular Bleeding, Softissue Haematoma and Psseudotumor In: -Merchan ECR, N.Goddard, Lee A, editors. Musculoskeletal Aspects of Haemophilia. 1st ed. Oxford: Wiley-Blackwell; 2000. p. 85-96.
11. Fernandez-Palazzi F, Hernandez SR, De Bosch NB et al. Hematomas within the iliopsoas muscles in hemophilic patients: the Latin American experience. Clin Orthop Relat Res. 1996;(328):19-24.
12. Arnold WD, Hilgartner MW. Hemophilic arthropathy. Current concepts of pathogenesis and management. J Bone Joint Surg Am. 1977;59(3): 287-305.
13. Shaheen S, Alasha E. Hemophilic pseudotumor of the distal parts of the radius and ulna. A case report. J Bone Joint Surg Am. 2005;87(11):2546-9.
14. Jelbert A, Vaidya S, Fotiadis N. Imaging and staging of haemophilic arthropathy. Clin Radiol. 2009;64(11):1119-28.
15. Rodriguez-Merchan EC. Musculoskeletal Complications of Hemophilia. HSS J. 2010;6(1):37-42.
16. Kim KY, Yang CH, Cho MJ et al.. Comprehensive clinical and statistical analysis of hemophilia in Korea. J Korean Med Sci. 1988;3(3):107-15.
17. Kar A, Potnis-Lele M. Descriptive epidemiology of haemophilia in Maharashtra, India. Haemophilia. 2001;7(6):561-7.
18. Chan V, Chan TK, Liu VW et al. Restriction fragment length polymorphisms associated with factor VIII:C gene in Chinese. Hum Genet. 1988;79(2): 128-31.
19. Manisha M, Ghosh K, Shetty S et al. Spectrum of inherited bleeding disorders from Western India. Haematologia (Budap). 2002;32(1):39-47.
20. Tonbary YA, Elashry R, Zaki Mel S. Descriptive epidemiology of hemophilia and other coagulation disorders in mansoura, egypt: retrospective analysis. Mediterr J Hematol Infect Dis.2(3.
21. Heim M, Rodriguez-Merchan EC, Horoszowski H. Orthopedic complications and Management of haemophilia. Inter J Paediatr Haematol Oncol. 1994;1:545-51.
22. Roosendaal G, Van den Berg HM. Blood Induced Joint Damage In: Rodriguez-Merchan E C GNj, Lee C A, editor. Muscloskeletal Aspect of Haemophilia. 1st ed. Oxford: Weily-Blackwell; 2000. p. 18-26.
23. Sajid R, Khalid S, Mazari N et al. Clinical audit of inherited bleeding disorders in a developing country. Indian J Pathol Microbiol. Jan-Mar;53(1):50-3.
24. Feil E, Bentley G, Rizza CR. Fracture management in patients with haemophilia. J Bone Joint Surg Br. 1974;56-B(4):643-9.
25. Biggs R. Thirty years of haemophilia treatment in Oxford. Br J Haematol. 1967;13(4):452-63.
26. Thomas HB. Some orthopaedic findings in ninety-eight cases of hemophilia. 1936. Clin Orthop Relat Res. 1997;(343):3-5.
27. Jordan HH. Hemophilic Arthropathies 1st US Edition ed.: Charles C Thomas, Springfield; 1958.
28. Mansouritorghabeh H, Rezaieyazdi Z, Saadati N et al. Reduced bone density in individuals with severe hemophilia B. Int J Rheum Dis. 2009;12(2):125-9.